CONDITION

Treacher collins syndrome

Treacher Collins syndrome is an inherited condition that affects how the face, jaw, ears, and related structures develop before birth.

WHAT IT IS
Genetic condition affecting facial bone and tissue development

BODY SYSTEM
Face, jaw, ears, eyes, and airway

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

WHAT CAUSES IT
Inherited or new genetic changes

HOW IT IS DIAGNOSED
Clinical examination and genetic testing

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

CAN IT BE CURED?
No - genetic cause cannot be removed

IS IT AN EMERGENCY?
Sometimes - serious breathing difficulty requires emergency care

LONG-TERM OUTLOOK
Usually normal lifespan with lifelong tailored care

Overview

Treacher Collins syndrome is a genetic condition that changes how parts of the face develop before birth. It mainly affects the cheekbones, lower jaw, ears, eyelids, and roof of the mouth. It is sometimes called a craniofacial condition, meaning a condition involving the skull and face.

The effects differ greatly from one person to another. Some people have subtle facial differences and mild hearing loss. Others are born with a very small jaw, significant hearing loss, feeding problems, or a narrow airway that makes breathing difficult.

Treacher Collins syndrome does not usually affect intelligence. However, untreated hearing loss, interrupted sleep, or long periods of illness can affect speech, learning, and wellbeing. Early hearing support and coordinated care can reduce these effects.

Care often involves several specialties over many years. The team may include hearing specialists, ear, nose and throat clinicians, craniofacial surgeons, dentists, orthodontists, eye specialists, speech therapists, dietitians, genetic specialists, and mental health professionals.

Symptoms

The signs are usually present at birth, although mild cases may not be recognised until later. Features often affect both sides of the face in a similar way.

Common features include:

A small jaw can leave less room for the tongue. The tongue may fall backward and partly block the airway, especially during sleep or in a newborn lying on their back. Possible signs include noisy breathing, pauses in breathing, poor sleep, daytime tiredness, or difficulty feeding.

Feeding may take longer because breathing, sucking, and swallowing need to be coordinated. Some babies struggle to gain weight. A cleft palate or severe jaw difference can add to these difficulties.

Eye symptoms can include dryness, irritation, or incomplete eyelid closure. This may happen because the lower eyelids do not fully protect the surface of the eye.

Hearing loss is often conductive. This means sound cannot pass normally through the outer or middle ear, even though the inner ear may work. Without suitable support, hearing loss can delay speech and language development.

- Underdeveloped cheekbones
- A small lower jaw and chin
- Eyes that slope downward toward the outer corners
- Notches or missing tissue in the lower eyelids
- Small, unusually shaped, or absent outer ears
- Narrow or missing ear canals
- Hearing loss caused by differences in the outer or middle ear
- A cleft palate, which is an opening in the roof of the mouth

Causes and risk factors

Treacher Collins syndrome is caused by a change in a gene involved in early facial

development. The genes most clearly linked to it include TCOF1 , POLR1B , POLR1C , and POLR1D .

These genes help cells make structures needed for producing proteins. During early development, certain genetic changes reduce the number of cells available to form the bones and tissues of the face.

Most cases linked to TCOF1 , POLR1B , or POLR1D follow an autosomal dominant inheritance pattern. This means one altered copy of the gene can cause the condition. A person with a dominant genetic change has a 1 in 2 chance of passing that change to each child.

Some cases linked to POLR1C , and some involving POLR1D , follow an autosomal recessive pattern. In this pattern, a child inherits an altered gene copy from both parents. The parents are usually healthy carriers.

The genetic change may be inherited from a parent, or it may occur for the first time in the egg, sperm, or early embryo. A new change is called a de novo variant. It is not caused by anything the parents did during pregnancy.

People in the same family can be affected very differently, even when they have the same genetic change. A mildly affected parent may have a child with more noticeable medical needs, or the reverse.

Diagnosis

Treacher Collins syndrome is often identified from its pattern of facial, jaw, ear, and eyelid features. A newborn with breathing or feeding problems may be assessed soon after birth.

The assessment may include:

Genetic testing can confirm the diagnosis when it finds a disease-causing change in a related gene. A negative genetic test does not always rule out the condition because current testing may not identify every possible cause.

A genetics professional can explain the result, the inheritance pattern, and what it may mean for relatives or future pregnancies.

If a familial genetic change is already known, prenatal genetic testing may be possible. Ultrasound can sometimes identify significant facial differences before birth, but it may not detect a mild case or predict exactly how severely a baby will be affected.

- Examination of the face, mouth, ears, and eyes
- Hearing tests suited to the person's age

- Imaging of the facial bones or airway when needed
- Sleep testing if sleep apnoea is suspected
- Feeding and swallowing assessment
- Genetic testing using a blood or saliva sample

Treatment and management

There is no treatment that removes the underlying genetic change. Care focuses on breathing, nutrition, hearing, communication, eye protection, dental health, and facial function.

Breathing has the highest priority in a newborn with a narrow airway. Support may include careful positioning, assisted breathing, or a device that keeps the airway open. Some children need surgery to lengthen the lower jaw or a temporary opening in the neck called a tracheostomy. The right approach is based on detailed airway assessment.

Feeding support may include changes in feeding position, specialised bottles, or help from a feeding therapist. Tube feeding may be used when eating by mouth is unsafe or does not provide enough nutrition.

Hearing should be assessed early and followed over time. Hearing aids that send sound through the skull bone may help when the ear canal or middle ear has not formed normally. Some people may later have ear reconstruction or another hearing procedure.

A cleft palate can be repaired surgically. Speech therapy may help with speech sounds, language development, or changes in voice caused by the palate and hearing loss.

Eye care may include lubricating drops or ointment and protection from wind or dust. Eyelid surgery may be considered when the eye surface is not adequately protected.

Facial and jaw surgery can improve breathing, chewing, eyelid function, or facial balance. Procedures may involve the jaw, cheekbones, ears, nose, or eyelids. Treatment is often staged because the face and teeth continue to grow. Not everyone wants or needs every available procedure.

Regular dental care is important because a small jaw can cause crowded teeth, an uneven bite, and difficulty cleaning some areas. Orthodontic treatment may be coordinated with jaw surgery.

Emotional support also matters. Visible differences, hearing loss, repeated operations, and other people's reactions can affect confidence and mental health. Counselling, peer support, and practical support at school or work may be useful.

Complications

A narrow airway can cause obstructive sleep apnoea. This means breathing repeatedly slows or stops during sleep because the airway closes. Untreated sleep apnoea can lead to poor sleep, daytime tiredness, behaviour changes, headaches, and strain on the heart in severe cases.

Other possible complications include:

Before an operation, the anaesthesia team needs to know about the condition and any previous airway difficulties. This allows extra planning for keeping the airway open during the procedure.

Complications are not inevitable. Regular hearing, airway, dental, eye, and growth checks can identify problems before they have a larger effect.

- Poor growth related to feeding difficulty
- Delayed speech or language from hearing loss
- Repeated ear problems
- Dental crowding and bite problems
- Eye irritation or damage from incomplete eyelid closure
- Stress, anxiety, low mood, or social isolation
- Difficulties with anaesthesia because of the small jaw and airway shape

Prevention

Treacher Collins syndrome cannot usually be prevented. It results from a genetic change that is inherited or occurs for the first time during early development. It is not caused by diet, exercise, stress, medication use without a specific known link, or a parent's actions.

Genetic counselling can help people understand their chance of passing on a known genetic change. Depending on personal circumstances and local availability, reproductive options may include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices rather than ways to prevent the condition in someone who already has it.

Early care cannot prevent the genetic condition, but it can reduce some secondary problems. Examples include supporting hearing and communication, monitoring breathing during sleep, protecting the eyes, and treating feeding or dental difficulties.

Living with it and what to expect

Treacher Collins syndrome is lifelong, but it is not progressive in the usual sense. The genetic condition does not keep damaging the body. Facial differences may become more or less noticeable as the face grows, and medical needs can change with age.

Infancy may involve the most urgent breathing and feeding decisions. During childhood, attention often shifts toward hearing, speech, dental development, sleep, schooling, and planned surgery. Teenagers and adults may consider further jaw, ear, or facial procedures, but these are based on function, personal goals, and informed choice.

Most people have typical intelligence and can attend mainstream education, work, have relationships, and live independently. Classroom seating, hearing technology, captioning, or other accommodations may make communication easier.

Many people have a lifespan similar to the general population, especially when significant airway problems are recognised and treated. The number of procedures and level of ongoing support differ widely.

Routine follow-up may include hearing tests, dental and orthodontic reviews, eye checks, growth monitoring, airway or sleep assessment, and review by a craniofacial team. Adults may need less frequent team care once facial growth and planned surgery are complete, but hearing, dental, sleep, and mental health needs can continue.

When to get medical help

Arrange a routine medical review for new or ongoing concerns such as:

Seek urgent medical advice if a baby or child is working harder than usual to breathe, repeatedly choking during feeds, having frequent pauses in breathing during sleep, or unable to take enough fluid.

Call emergency services for severe breathing difficulty, blue or grey lips or skin, loss of consciousness, or a prolonged pause in breathing. These signs need immediate assessment.

- Snoring, restless sleep, morning headaches, or daytime sleepiness
- Hearing changes or difficulty following conversation
- Feeding that regularly takes a long time
- Poor weight gain
- Eye dryness, redness, or discomfort

- Dental pain, crowding, or difficulty chewing
- Speech or language concerns
- Anxiety, low mood, bullying, or withdrawal from usual activities

Common questions

Does Treacher Collins syndrome affect intelligence?

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

Does Treacher Collins syndrome get worse with age?

It is not generally progressive. The underlying condition does not spread or cause continuing tissue damage. However, growth can change facial proportions, dental alignment, breathing, and the timing of treatment.

Can surgery cure Treacher Collins syndrome?

No. Surgery cannot remove the genetic cause. It can improve specific concerns such as airway narrowing, cleft palate, eyelid protection, chewing, hearing access, or jaw function.

Can someone with Treacher Collins syndrome hear normally?

Some people have normal or near-normal hearing, but conductive hearing loss is common. Hearing devices or surgery may improve access to sound. Regular testing is important because a young child may not be able to describe hearing difficulty.

Can I pass Treacher Collins syndrome to my children?

It can be passed on, but the chance depends on the gene and inheritance pattern. With a dominant genetic change, each child has a 1 in 2 chance of inheriting it. A genetics professional can interpret a specific family result.

What is the life expectancy for Treacher Collins syndrome?

Life expectancy is usually similar to that of the general population. Severe airway problems can be dangerous, especially in infancy, but careful airway assessment and treatment greatly influence the outlook.

Sources



- GeneReviews: Treacher Collins Syndrome , NCBI Bookshelf, University of Washington, Seattle
- Treacher Collins syndrome , MedlinePlus Genetics, U.S. National Library of Medicine
- Treacher Collins syndrome , NHS
- Treacher Collins Syndrome , National Organization for Rare Disorders
- Treacher Collins syndrome , Orphanet
- Treacher Collins Syndrome , Seattle Children's Hospital