



Treacher Collins Syndrome

MANDIBULOFACIAL DYSOSTOSIS

2026 Edition

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What Is Treacher Collins Syndrome?

Treacher Collins syndrome (TCS) is a rare group of facial differences that are present at birth. There are currently about 10,000 people in the U.S. who were born with TCS. The syndrome affects one in 50,000 births. TCS, also called mandibulofacial dysostosis or Franceschetti syndrome, affects the growth of the face's bones and tissues. The severity of the syndrome varies greatly from person to person. TCS affects the size, shape, and position of the ears, eyes, eyelids, cheekbones, and jaws.

What Causes TCS?

During pregnancy, there are many genes involved in the development of the face. TCS occurs when one of these genes changes (called a mutation). Scientists have identified three genes (TCOF1, POLR1C, and POLR1D) linked to TCS, though others remain unknown. In about half of cases, TCS is passed down genetically from a parent to a child (inherited). In other cases, the mutation happens randomly, without a known cause. A geneticist is a doctor who studies how conditions like TCS are passed down in families. A geneticist can diagnose TCS by examining a child's facial features. The geneticist can also order blood tests to help families learn more about the genes involved.

If I Have Other Children, Will They Have TCS?

Every person carries two copies of a gene. When two people have a child, the child inherits one copy of a gene from each parent. If one parent has a mutated gene, the child has a 50% chance of inheriting it. With TCS (and other conditions that follow this pattern), the child will be born with the syndrome if he or she inherits that gene. This genetic pattern is called autosomal dominant.

If one parent was born with TCS, the chances of having a child with the syndrome are 50% (one out of two). If neither parent was born with TCS, the chances of having a second child with TCS are the same as they were for their first child, .0012%, or about 1 chance in 83,000. In other words, the chances are about 1 in 83,000 that a gene related to TCS will mutate randomly, causing the syndrome.

A geneticist can help determine if a child with TCS has inherited the condition from a parent or if it happened by chance. This process usually involves blood tests for mutations in the genes listed above. If you are pregnant and wonder whether you will have a child born with TCS, prenatal counseling can help you learn more. Severe forms of TCS can be seen on a prenatal ultrasound.

What Are the Facial Differences of TCS and How Are They Treated?

TCS can affect many parts of the face. However, the extent of facial differences varies from person to person. A person with TCS may have some of the characteristics listed below, but not others. Also, any one of these characteristics, if present, could be mild or severe.

THE AIRWAY AND JAW

Several features of TCS can cause problems with the airway and breathing:

- A narrow airway in the nose, called choanal atresia. Choanal atresia occurs in 11% of people born with TCS. This nasal obstruction is caused by extra bone or tissue in the back of the nose.
- A drooping of the tongue, called glossoptosis.
- Problems with the voice box or lower airway.
- A set-back upper jaw (maxilla), with steeply angled teeth. When the maxilla is set back, the upper and lower teeth may not come together properly, causing a gap between the upper and lower front teeth (an anterior open bite). In some cases, the position of the upper jaw can cause breathing problems.
- A small lower jaw (mandible) that is not normally shaped. This condition is called micrognathia or retrognathia. As with the upper jaw, the lower jaw may be set back and steeply angled. Later in childhood, the teeth may not align properly. These problems with the size and shape of the lower jaw can limit how the mouth opens and can cause early problems with breathing and eating. For 25-40% of people with TCS, this condition can cause sleep apnea, a sleep disorder related to breathing.

Some babies with TCS have mild breathing problems that can improve with position changes during sleep. For example, breathing may improve when they lie on their side or belly instead of their back. Make these changes with a doctor's advice and monitor them. In mild cases, a doctor may insert a temporary tube called a nasal trumpet to open the airway. Another option is to wear a special breathing mask called CPAP (*Continuous Positive Airway Pressure*).

If problems with breathing are severe, a few treatment options may be offered:

- An endotracheal tube is a breathing tube that goes into the airway through the nose or mouth to help with breathing. This tube goes deep into the airway, past the tongue and vocal cords. An endotracheal tube prevents a person from eating through the mouth. This tube is a temporary solution to help with breathing (it cannot be used long-term).



Figure 1: If a person born with TCS has a small or abnormally shaped lower jaw, he or she may also have a small airway. In some cases, a small airway can cause sleep apnea or other breathing problems.



Figure 2: Distraction osteogenesis is a procedure to lengthen the jaw. After cutting the jawbones, a surgeon places a device called a distractor. The distractor lengthens the bones by slowly separating them.

- A tracheostomy is a breathing tube placed into the neck by a surgeon. This tube can be temporary or permanent.
- If a child has a narrow airway in the nose (called choanal atresia), a surgeon can open up the space by removing extra tissue in the back of the nose.
- Jaw lengthening, called distraction osteogenesis, begins with an operation to cut the jawbones and place a device called a distractor. The distractor then slowly separates the bones to lengthen them. Lengthening the jaw can help with breathing. It can also help a person avoid a tracheostomy. In some cases, if a tracheostomy is already in place, jaw lengthening can improve the airway enough for its removal.
- Additional reconstructive jaw surgery may be needed during the teenage or adult years to correct the bite and treat airway obstruction (for more information on craniofacial reconstruction, see the section on the cheekbones and jaw, below).

- Many people with TCS have frequent sleep studies. Breathing issues can return, even after treatment. Breathing must be monitored throughout life.

FEEDING AND GROWTH

Many babies and children with TCS have trouble eating. These difficulties usually occur with a cleft palate or breathing problems. In other cases, a person with TCS may have trouble eating because the opening in the mouth is small or because the top and bottom teeth don't meet the way they should. Some people with TCS have problems with the salivary glands. The salivary glands produce liquid (saliva) to keep the mouth moist. If these glands don't produce enough saliva, a person can have trouble chewing and swallowing food.

A baby with TCS may need help from a feeding specialist. He or she may also require a feeding tube. With time, most children with TCS eat normally. The craniofacial team monitors eating and weight during childhood.

THE PALATE

Some people born with TCS have a high arch in the roof of the mouth. Of all people born with TCS, 28% have either a hole in the roof of the mouth (called a cleft palate) or a gap between the muscles under the mucous membrane of the roof of the mouth (called a submucous cleft palate). In most cases, a surgeon closes a cleft palate during a baby's first year. Closing a cleft palate helps with feeding and speech (see the ACPA booklet, "Preparing for Surgery," to learn more). Occasionally, closing a cleft palate increases the risk of airway obstruction. A baby with a submucous cleft palate may not need surgery right away. The team usually repairs this type of cleft later in childhood if it affects speech (see the ACPA booklet, "Submucous Cleft Palate," to learn more).

THE EYES

Many people born with TCS lack bone and tissue around the eyes. As a result, the eyes may slant downward. Each lower eyelid may also have a notch called a coloboma. Eyelashes may also be missing on the inner edges of the lower eyelids. Sometimes, a lack of eyelid tissue can cause eye dryness. Although blindness is not part of TCS, dryness can damage the eyes and cause some vision loss. The craniofacial team usually suggests operating on parts of the eyes and eyelids early in life. The team may also suggest other procedures at different times in childhood, for appearance or to protect the eye.



Figure 3: A person with TCS may have eyelids that slant downward. They may also have a notch or gap in the lower eyelids called an eyelid coloboma. Underdeveloped or missing cheekbones and abnormally shaped eye sockets (orbits) contribute to this appearance.

THE CHEEKBONES AND JAW

When a person is born with TCS, parts of the cheekbones may be missing or small. These differences can affect cheek shape and eye position. A craniofacial team has many ways to build and change parts of the face. Using a procedure known as bone grafting, a surgeon can transplant bone from one area of the body to another. Bone grafts may come from the skull (calvarial bone) or the ribs. Bone grafts and synthetic implants can build cheekbones.

Corrective jaw surgery, called orthognathic surgery, corrects the bite, changes the appearance of the face, reduces mouth breathing, decreases airway obstruction, and improves a person's ability to chew. If the jaw joint (temporomandibular joint, or TMJ) is not formed correctly, new jaw joints (ball-and-socket) may also be needed. These joints can be rebuilt using bone grafts from the ribs or from man-made materials (for more information, see the ACPA publication, "As You Get Older").

The methods and the timing of cheekbone and jaw procedures depend on a child's physical symptoms, breathing, and social and emotional health. They also depend on the team's experience. This type of work is called craniofacial reconstruction. It usually happens during the teenage years.

THE EXTERNAL EAR

Many people with TCS are born with a small or missing external ear, the part of the ear that shows when you look at a person. This condition is called microtia. A surgeon can build an ear using tissue from somewhere else in the child's body (usually from the ribs) over the course of two or three operations. Another option is to build an ear in just one operation using synthetic material called auricular construction.



Figure 4: Microtia is a condition where the external ear is small, abnormally shaped, or missing.

In some cases, a surgeon may suggest an artificial ear (also called a prosthetic ear). Prosthetic ears are made of synthetic material to look like an external ear. They are held in place with special glue or with an implant. Building an outer ear does not affect hearing.

HEARING AND SPEECH

If a person with TCS is born with a small or missing external ear, he or she may also have irregularities inside the ear. Some people with TCS have a narrow or missing ear canal, called aural atresia, which can cause hearing loss. A person with TCS may also lack an eardrum or middle ear bones (called ossicles). In some cases, surgeons can rebuild these structures to restore hearing.

In other cases, the ear canal, eardrum, and middle ear bones are present at birth but irregularly shaped. These differences usually cause conductive hearing loss on one or both sides. With this type of hearing loss, the hearing nerve works normally, but sound cannot reach it through the middle-ear structures. A traditional hearing aid rarely helps a person with TCS. A Bone-Anchored Implant or Bone-Anchored Hearing Aid (BAI or BAHA), however, can help transmit the sound. A headband holds this device in place for a baby or younger child. After age 4, it can be placed surgically into the skull behind the ear. Experts are currently developing a product that combines a BAHA with a prosthetic ear.

Many people with TCS also have fluid in the ears, called chronic otitis media. This problem is common for people born with a cleft palate. Pressure equalization tubes (commonly called ear tubes) are tiny tubes a surgeon places in the eardrum. By allowing the fluid to drain, ear tubes can improve hearing and reduce the chances of ear infections.

People with hearing problems usually have a hard time learning to speak. Fortunately, normal speech is possible for people with TCS once they can hear properly.

Children born with a cleft palate may develop speech issues, such as a nasal tone of voice. After the cleft palate is repaired, children should have regular speech evaluations from the speech-language pathologist on the team. Sometimes, another operation is needed to correct speech issues. In many cases, though, speech therapy can solve speech problems.

THE NOSE

About one in 10 people with TCS (11%) are born with extra bone or tissue in the back of the nose, called choanal atresia or stenosis (as mentioned previously). This extra material narrows the airway, making breathing difficult. Surgery can open up the space. If the wall between the nostrils is curved or crooked, called a deviated septum, another procedure can straighten it. Both procedures can help with breathing.

People with TCS often have a curved bridge of the nose. The nose may look prominent because the jaw and cheekbones are small. Later in life, a child may have an operation to change the nose's shape and appearance (the combined procedures are called septoplasty and rhinoplasty). The team usually recommends doing this operation later in adolescence, following craniofacial reconstruction (described above).

SOFT TISSUES

For some people with TCS, the skin and fat of the cheeks and temples, called facial soft tissues, are thin. The cheek muscle may also be separated from the tissues. This separation can cause a dent in the cheeks, near the corners of the mouth. Later in childhood, after craniofacial reconstruction, the team may suggest ways to build up the cheek tissues. One method, fat grafting, involves injecting fat removed from elsewhere in the body (using liposuction) into the cheeks. Another approach, an operation called a free flap, involves transferring larger pieces of tissue that include blood vessels. Both procedures are called soft tissue augmentation.

Will Facial Differences Change Over Time?

The facial differences associated with TCS are not progressive, meaning that they do not worsen over time. As a person grows, however, parts of the face will get larger and change in appearance. The size and shape of the airway will also change over time. The health professionals on the craniofacial team should monitor your child's growth and development regularly during childhood (including vision, hearing, sleeping, and oral health).

Will My Child Be Developmentally Delayed?

Most children with TCS have normal ability to think and reason (called cognitive development). Intelligence is usually normal and, in some cases, higher than the general population. Some children with TCS have learning delays. Hearing loss and a cleft palate, for instance, can interfere with the process of learning speech. In many cases, a speech delay can be shortened or avoided with early testing and treatment.

What Is the Timeline for Treatment?

Each person with TCS will have a different treatment plan. The sample timeline below lists a common sequence of treatments for TCS—but it is just one example. A plan depends on the child's needs, the team's approach, and the wishes of the child and family. The timing also depends on a person's development. Some treatments can only be performed early in life. Others are effective in the teenage years, after a child grows.

EARLY INFANCY

Some babies with TCS need an operation shortly after birth to help with breathing, such as tracheostomy or mandibular distraction osteogenesis. In some cases, surgery is needed to treat a blockage in the nose (choanal atresia) to help with breathing.

FIRST YEAR

During the first year of life, some babies will have surgery to close a cleft palate. If needed, they may also place ear tubes. If a baby has a notch in the eyelid (a coloboma), the team may recommend an operation to repair it. A baby with TCS may wear a headband-style hearing aid during this time.

CHILDHOOD

At age 4 and older, a child can receive a bone-anchored hearing aid (BAHA). Sometimes, tonsils need to be removed at this time to improve breathing or address infections. After age 6, a team may recommend surgery to build the ear.

TEEN YEARS

After a child is done growing, the team may recommend craniofacial reconstruction. This phase of treatment may include operations on the lower jaw (mandible), upper jaw (maxilla), eye sockets (orbits), cheekbones, and/or nose. After craniofacial reconstruction, a child may undergo soft tissue augmentation and rhinoplasty.

EVERY YEAR

A child with TCS should have regular checkups for breathing and sleeping. The team may recommend sleep tests, as needed. Also, if a child was born with a cleft palate or submucous cleft palate, he or she should have speech evaluations starting at age 2 or 3. Every child with TCS should visit the craniofacial team every year.

Where Can I Find Support?

Every parent will have his or her own responses to learning about a child's TCS. As you work through your feelings and learn more about the condition, consider seeking emotional support. Some families feel reassured when they talk with others going through the same experience. Some find comfort through counseling or an online support group. There are many options.

If you have just learned about your child's TCS at birth or have learned the news through prenatal diagnosis, be sure to discuss your concerns with your craniofacial team. The team will:

- Work with parents to make and carry out a comprehensive treatment plan for a child
- Provide education and support related to feeding and breathing
- Connect parents and children to sources of emotional support, including other families with children born with TCS
- Teach families about hearing and speech
- Offer support before and after operations

To learn more about choosing a team, see the ACPA Factsheet, "Information About Choosing a Cleft Palate/Craniofacial Team." The team should be a family's main resource throughout childhood for care, planning, and support.

Glossary

AUDIOLOGIST - A medical professional who specializes in hearing and balance.

AURAL ATRESIA - A condition in which the ear canal is narrow, closed, or missing. Aural atresia can cause hearing loss.

BAHA (BONE-ANCHORED HEARING AID, ALSO CALLED BONE-ANCHORED IMPLANT, OR BAI) - A type of hearing aid that transmits sound through bone. This type of hearing aid can be held against the head with a headband or surgically attached to the skull.

CHOANAL ATRESIA - A condition in which the back of the airway in the nose (the nasal passage) is blocked by bone or soft tissue.

CLEFT - A hole sometimes found in the lip or the roof of the mouth (palate). A cleft lip and/or palate occurs between weeks 5 and 12 of pregnancy.

COLOBOMA - A gap in the structures of the eye. People with TCS often have a coloboma of the lower eyelid.

CPAP THERAPY (CONTINUOUS POSITIVE AIRWAY PRESSURE) - A method to help with breathing in which a person receives air through a mask.

CRANIOFACIAL SURGEON - A surgeon who specializes in building and rebuilding parts of the head and face.

DISTRACTION OSTEOGENESIS - A process of lengthening a bone. A surgeon cuts the bone and inserts a device called a distractor, which slowly moves the two pieces of bone apart. The bone heals in between the space and, over time, gets longer.

EXTERNAL EAR - The outer portion of the ear that you can see when you look at someone.

FREE FLAP - An operation in which a piece of tissue, including its blood vessels (a vein and an artery), is removed from one part of the body and connected to new blood vessels in a different part of the body.

GENETICIST - A doctor who understands how conditions are passed down in families. A geneticist studies genes, heredity, and family traits.

GLOSSOPTOSIS - A backward or downward drooping of the tongue that can block the airway and make breathing difficult.

GRAFT - Tissue moved from one area of the body to another that develops a new blood supply in its new location.

INTERNAL EAR - The part of the ear inside the skull. The internal ear contains the structures involved in hearing.

INTUBATION - Placing a temporary breathing tube in the airway to help with breathing.

MANDIBLE - The lower jaw.

MAXILLA - The upper jaw.

MEDPOR® (ALSO CALLED POROUS POLYETHYLENE) - A type of synthetic material that can be used to build or rebuild parts of the face, such as ear cartilage or cheekbones.

MICROGNATHIA - A small lower jaw.

MICROTIA - A small or absent external ear. People with microtia often have problems with inner-ear structures as well.

MUTATION - A change in the coding of a gene.

OBSTRUCTIVE SLEEP APNEA - A condition where breathing is partly or completely blocked, causing shallow breathing or pauses in breathing during sleep. This condition is sometimes called “sleep-disordered breathing.”

ORAL/MAXILLOFACIAL SURGEON - A surgeon with dental training who specializes in building and rebuilding the mouth, jaws, and face.

ORTHOGNATHIC SURGERY - An operation, also called corrective jaw surgery, in which the jawbones are adjusted and constructed to correct problems with the bite.

OSSICLES - The tiny bones in the middle ear that carry sound vibrations to the inner ear, allowing hearing.

OTOLARYNGOLOGIST (ENT OR EAR, NOSE, AND THROAT DOCTOR) - A surgeon who specializes in conditions that affect the ears, nose, airway, swallowing, and speech.

PALATE - The roof of the mouth. The palate separates the mouth from the nose and helps with eating and speech.

PLASTIC SURGEON - A surgeon who builds and rebuilds parts of the body to change function or appearance.

PROSTHODONTIST - A dental specialist who designs, makes, and fits devices — such as an obturator — to replace or support missing or misshapen parts of the mouth and face.

RETROGNATHIA - A condition in which the lower jaw is set back (retrusive).

RHINOPLASTY (ALSO CALLED NOSE JOB) - An operation that changes the shape of the nose and helps improve nasal breathing.

SEPTOPLASTY - An operation that straightens the wall between the nostrils (the septum) to improve breathing. Septoplasty is often performed along with rhinoplasty.

SLEEP STUDY (ALSO CALLED POLYSOMNOGRAM) - A test of how well a person breathes during sleep and helps identify sleep apnea.

SUBMUCOUS CLEFT PALATE - A minor form of cleft palate in which the lining of the palate is intact but the muscles beneath it are separated. Many children with this condition have a split uvula, a division in the tissue that hangs at the back of the palate.

TRACHEOSTOMY - A breathing tube that is surgically placed through the neck.



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FOR MORE INFORMATION

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