



TREACHER COLLINS SYNDROME

MANDIBULOFACIAL DYSOSTOSIS



ACPA Family Services
Resources for your cleft journey

TREACHER COLLINS
SYNDROME
MANDIBULOFACIAL
DYSOSTOSIS

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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 bone and tissues of the face. The severity of the syndrome varies greatly from one person to the next. 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 (known as TCOF1, POLR1C, and POLR1D) that relate to TCS, though there are others still unknown. In about half of cases, TCS is passed down genetically to a child from a parent (*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 of the parents 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. The extent of the facial differences, however, varies from one person to another. 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

There are several features of TCS that 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 voicebox 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 as they should, causing a gap between the upper and lower front teeth (called an *anterior open bite*). In some cases, the position of the upper jaw causes problems with breathing.

- 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.



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 problems with breathing.

Some babies with TCS have mild breathing problems that can be helped by changing position during sleep. Breathing may be improved by lying on the side, for instance, or by lying on the belly instead of the back. These kinds of changes must be made with the advice of a doctor and must be monitored. In mild cases, a doctor may insert a temporary tube called a *nasal trumpet* into the nose 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 as a permanent solution).
- 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 sur-

geon 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 lengthens the bones by separating them slowly. 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.



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 separating them slowly.

- 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 construction*, see the section on the cheekbones and jaw, below).
- Many people with TCS have frequent sleep studies. Breathing issues can come back, even after certain problems have already been treated. Breathing must be watched throughout life.

FEEDING AND GROWTH

Many babies and children with TCS have trouble eating. These difficulties usually occur if a person has a cleft palate or has problems with breathing. 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 kids 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 problems with 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 be missing on the inner edges of the lower eyelids as well.

Sometimes, a lack of tissue in the eyelids can cause dryness of the eye. Although blindness is not part of TCS, dryness can lead to damage to the eyes and some loss of sight. 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.



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Figure 3: A person with TCS may have eyelids that slant downward. He or she may also have a notch or gap in the lower eyelids called an eyelid coloboma. Underdeveloped or missing cheekbones and abnormally shaped eye sock-ets (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 the shape of the cheeks

and the position of the eyes. 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 be taken from the skull (known as *calvarial bone*) or from the ribs. Bone grafts and synthetic implants can be used to build cheekbones.

Corrective jaw surgery, called *orthognathic surgery*, is a procedure to correct the bite, change the appearance of the face, reduce mouth breathing, decrease airway obstruction, and improve a person's ability to chew. If the jaw joint (*temporomandibular joint* or *TMJ*) is not formed correctly, it is possible that new jaw joints (*ball and socket*) will be needed as well. These joints can be rebuilt using bone grafts from the ribs or from man-made materials (for more information, see the ACPA booklet, "Information for Teens").

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 experience of the team. This type of work is called *craniofacial construction*. 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



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

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 *Medpor*. The process of building an ear is called *auricular construction*.

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 has no impact on 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. It is also possible that a person with TCS lacks an eardrum or middle ear bones (called *ossicles*). In some cases, these structures can be built through surgery, making hearing possible.

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 the sound cannot reach it through the structures of the middle ear. 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. This device is held in place by a headband 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 placed into the eardrum by a surgeon. 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 issues with speech, 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 aspects of speech. In many cases, though, speech problems can be solved with speech therapy.

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, causing difficulties with

breathing. 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.

It is common for people with TCS to have a curved bridge of the nose. The nose may look prominent because of the small size of the jaw and cheekbones. Later in life, a child may have an operation to change the shape and appearance of the nose (the combined procedures are called *septoplasty* and *rhinoplasty*). The team usually recommends doing this operation later in adolescence, following *craniofacial construction* (described above).

SOFT TISSUES

For some people with TCS, the skin and fat of the cheeks and temples, called *facial soft tissues*, are thin. Also, the muscle of the cheek may 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 construction*, the team may suggest ways to build up the tissues of the cheeks. One method, *fat grafting*, involves injecting fat into the cheeks that has been removed from elsewhere in the body (using *liposuction*). Another approach, an operation called *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.

WHEN 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 approach of the team, 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, ear tubes will be placed as well. 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 help with breathing or solve problems with 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 construction*. This phase of treatment may include operations on the lower jaw (*mandible*), upper jaw (*maxilla*), eye sockets (*orbits*), cheekbones, and/or nose. After craniofacial construction is finished, 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 find out more about the condition, consider looking for 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.

BAHA: 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 attached to the skull surgically.

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.

Graft: Tissue that is moved from one area of the body to another and 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: The placement of 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 structures of the inner ear 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 referred to as “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.

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.

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.

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.

FOR MORE INFORMATION

This booklet and many others have been produced by:

The American Cleft Palate-Craniofacial Association

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MORE INFORMATION FOR YOUR FAMILY

ACPA Family Services has information for every step of your journey. We work with cleft and craniofacial care professionals to deliver the best information when you need it most.

“When we found out Vivienne had a cleft, we were scared of what her future would hold. There were days that felt so bleak. However, we overcame that – I read many “success stories” like the ones on the ACPA website, I consulted the family resources page on www.cleftline.org, I became my child’s best advocate, I took the advice that helped and ignored the rest, I surrounded myself with positive people as much as possible, and I believed in the best for Vivienne and our family.”

-Kristin

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