a guide to understanding

treacher collins syndrome

a publication of children's craniofacial association

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this guide to Treacher Collins syndrome is designed to answer questions that are frequently asked by parents of a child with Treacher Collins syndrome. It is intended to provide a clearer understanding of the condition for patients, parents and others.

how can children's craniofacial association (cca) benefit my family?

CCA provides programs and services that address the medical, financial, psychosocial, emotional, and educational concerns relating to craniofacial conditions. We offer financial assistance for medical travel, family networking to talk to people who have had similar experiences, gatherings to connect children and caregivers, and educational materials to help your family advocate to classrooms, groups, and the general public. For the most current updates on our programs, please visit our website at www.ccakids.org. CCA understands that when one family member has a craniofacial condition, each person in the family is affected. We provide programs and services designed to address these needs. A detailed list of CCA's programs and services may be found on our website at **www.ccakids.org** or call us at **800.535.3643.**

The information provided here was written by a member of the Medical Advisory Board of Children's Craniofacial Association, with the help of Michael L. Cunningham, MD, PHD in 2019 and updated in 2026.

This booklet is intended for information purposes only. It is not a recommendation for treatment. Decisions for treatment should be based on mutual agreement with the craniofacial team. Possible complications should be discussed with the physician prior to and throughout treatment.

Design by Khadija Moten

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what is treacher collins syndrome?

treacher Collins syndrome is a genetic condition in which the the facial bones and ears are underdeveloped. It is also called mandibulofacial dysostosis. This condition was named after a British ophthalmologist, Dr. Treacher Collins, who in 1900, described two children with very small cheekbones and notches in their lower eyelids. Children with Treacher Collins syndrome have small and differently shaped jaws, small cheek bones, small or absent ears, and notching lower eyelids.

why does treacher collins syndrome happen?

there are two ways that Treacher Collins syndrome develops. First, Treacher Collins can develop as a new genetic change. This means neither parent has the Teacher Collins mutation but it occurs at the time of conception. There is no evidence that the mother's actions or activities during her pregnancy contribute to this condition.

The second way that Treacher Collins syndrome develops is by inheriting it from one of the parents. It should be noted that sometimes one of the parents may have such a mild form of the condition that it goes undetected. Sometimes it is not until the birth of a child with Treacher Collins that it is determined that one of the parents has the mutation. Genetic testing and genetic counseling can help families understand their genetic outlook and is usually a part of the diagnostic process.

what are the chances of having a child with this syndrome?

It is estimated that Treacher Collins syndrome occurs in one of 1/50,000 births. In most cases, if neither parent has a mutation their chance of having a second child with Treacher Collins does not change (1/50,000). The reason we say "in most cases" is because on rare occasions only the mothers eggs or the fathers sperm have the mutation and this can be missed in routine mutational analysis of blood)

Adults with Treacher Collins syndrome have a 50% chance of passing the condition to their offspring. When a parent with Treacher Collins syndrome passes on the mutation, the children may be affected in varying degrees. The degree may be the same as the parent, milder, or more severe. Genetic testing and genetic counseling can help individuals understand their own genetic outlook and can be a part of craniofacial team visits as the child ages.

CCA encourages families to research teams on the list of approved teams maintained by American Cleft-Palate Craniofacial Association (ACPA), found at: https://acpacares.org/find-a-care-team .

what medical concerns are common in people with treacher collins syndrome?

Several concerns are common to Treacher Collins syndrome. A child does not necessarily have all of these concerns. The most common difficulties involve **breathing**, **hearing**, and **vision**.

BREATHING

Children with Treacher Collins syndrome have small underdeveloped jaws. This can cause the tongue to be positioned farther back in the throat, resulting in a smaller airway. Another common concern may be a blockage of their nasal airway, known as choanal atresia. The airways can become even narrower when children develop colds because of congestion and swelling.

when does breathing become a concern?

If your child with Treacher Collins syndrome has symptoms of breathing problems, like snoring or increased breathing effort, it is important to see your physician, because your child may need a sleep study to make sure that he or she is not developing sleep apnea. **Sleep apnea** is a condition in which the child is not breathing properly during sleep. The child may even stop breathing for a time. It is now believed that sleep apnea may affect the child's development.

what can be done to prevent breathing concernns?

Some children with a severe form of Treacher Collins require **tracheotomies (trachs)** early in life. In addition, some children will have an abnormality of the palate. The **palate** is another name for the roof of the mouth. Children who have a cleft palate will need corrective surgeries. They may also need speech therapy.

what are other effects of breathing concerns?

Children with severe breathing problems may have difficulties with feeding. If there is difficulty with breathing, the child often requires more calories in the diet. Since it is impossible to swallow and breathe at the same time, these children may not be able to eat an enough food on their own for adequate growth. Therefore, they may require a **gastric tube (g-tube)** for supplemental feeding. Finally, breathing problems over a long time can eventually affect the heart. These conditions require close monitoring by a pediatrician.

is there surgical correction for the jaw?

Surgery to lengthen the jaw usually occurs in stages. The timing depends on the how much the jaw is affected. The more underdeveloped the jaw, the earlier a surgery may need to be performed to lengthen the bone in order to improve breathing, feeding, and appearance. The surgeries recommended depend on the severity of jaw involvement and the treating center but may include bone grafting or a technique called **distraction osteogenesis,** which uses a a temporarily-implanted expanding device to gradually lengthen the bone.

HEARING

Most children with Treacher Collins have an abnormality of the external ear, which is called **microtia**. These differences range from a prominent ear to a complete absence of the external ear. They may have ears that prevent sound from entering the ear or abnormalities with how the ear processes sound.

do children with treacher collins have hearing loss?

It is important to have your child's hearing tested right after birth. Most hospitals do this before your are sent home. Although it is possible for children to hear without an outside opening in the ear, the sound will be quite muffled. If this is the case, it is very important to get a hearing aid in the first few months of life.

why is it important to get a hearing aid before the age of six months?

The sounds that babies hear in the first year are very important for language development. It was previously believed that most children with Treacher Collins had developmental delays. It is now realized that they have the same ability to learn as other children, when given hearing aids, so they can hear properly in school and other environments.

ear reconstruction

Reconstruction of the ears can vary greatly, but typically happens in the "early elementary" age range. See our booklet on Microtia for more information about ear reconstruction. External ears can be built from the child's own rib cartilage, made from a mixture of artificial material and the child's own tissue, or can be made as prosthetic attachments.

If the child is a candidate for ear canal and middle ear reconstruction, it may be coordinated with external ear surgery. Some centers operate on both ears at once to decrease the number of surgeries for the child. Other children may be candidates for hearing aid implants. It is important to coordinate surgical ear reconstruction with any bone-anchored hearing aid surgeries that are planned.

VISION

The eyes can be one of the more noticeable features of Treacher Collins. A child with this syndrome may have eyelids that can droop or have notches in them. If desired, surgery is available to partially correct the appearance. It is very important that patients with Treacher Collins syndrome are followed by an ophthalmologist. Eye ointments may need to be used at night to prevent the eyes from drying out which can lead to eye injury.

when is the best time to look in to treatment?

The best time to look into treatment is right after birth with a referral to a craniofacial team. Working with a team will allow you to make a treatment plan, even if you will not begin treatment for years. The timing for different surgical procedures varies among surgical treatment centers and according to the severity of the conditions. Surgery to rebuild the cheekbones usually begins before the child reaches the age of five. At the time of this surgery, the outer corners of the eyes may be raised to tighten the lower eyelids.

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mental health

Children and adults may face mental health struggles when living with a facial difference. CCA advocates that parents try a multifaceted approach to creating mental resiliency, including counseling/therapy, social support groups/networking, and encouraging self-care for the entire family.



children's craniofacial association

to support and inspire individuals and families affected by facial differences

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