

Information for Parents

TREACHER COLLINS SYNDROME (Mandibulofacial Dysostosis)

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Clinical Features

Treacher Collins Syndrome (TCS) is a *genetic* disorder that affects the eyelids, cheekbones, ears, and jaws. Intelligence is normal. TCS can be confused with the more common non-genetic condition, hemifacial microsomia. Two distinguishing features of TCS:

1. It is remarkably symmetrical
2. There is no involvement of the facial nerve (weakness of the facial muscles)

There is a wide spectrum of features (*expressivity*), meaning some children in a particular family are mildly affected and others more severely affected. Typically, there is a downward slant of the corner of the eyes. In 75% of children, a notch (*coloboma*) occurs in the lower eyelids at the junction of the outer and middle third, sometimes in the upper eyelid as well. There is almost always absence of lashes along the medial one-third of the lower eyelid. The tear drainage system of the lower eyelid can be affected. The eyes are usually normal; there are rare cases of small eyes or a slit layer of the eyes. The external ears are often small, malformed, and low set in position (*microtia*). Children often have a conductive hearing loss associated with abnormalities of the internal and middle ear. There is often a characteristic extension of temporal hair ("side burns") onto the cheek in front of the small ears. The nose tends to be relatively broad, large and beak-like. The *zygomas* (cheekbones) are undeveloped; they may be almost totally absent.

An infant with TCS can have trouble breathing in the neonatal period. This is due to the underdevelopment of the lower jaw (so that it is set back) and the tongue falls backward, narrowing the airway. The dimensions of the pharynx and nasopharynx are about one-half of normal size. Additionally, these children often have narrowing of the *choanae*. The palate is high arched; a cleft palate is present in about 30% of children.

Etiology

TCS occurs in a frequency of about 1 in 10,000 live births. It is inherited as an *autosomal dominant trait* with variable *penetrance* and *expressivity*. There is no conclusive family history of the condition in about 50% of affected children; this means there is a high rate of spontaneous *mutation* to the abnormal *gene*. TCS is caused by a mutation on the long arm of *chromosome 5*. The mechanism by which this chromosomal change produces the abnormal facial features is under active investigation.

Evaluation

Evaluation for an adequate airway is critical in the newborn nursery. The hearing should be evaluated in the first three months, with a brain stem evoked potential study. Children should be followed from infancy by an experienced craniofacial team.

Management

From the time of birth, the airway must be secured. Usually proper attention to position and feeding the infant is all that is necessary. Some severely affected infants with TCS require semi-emergent tracheostomy.

In order for development of normal language, periodic evaluations by an audiologist, otolaryngologist, and speech pathologist are imperative. A conductive hearing aid may be needed to enhance audiologic stimulation.

During the first year of life, small surgical procedures may be indicated, such as closure of notching of the eyelids and repair of a cleft palate. The more major craniofacial abnormalities are addressed in childhood. Anesthesia for these children is critical and special attention must be given to management of the airway, both during and after an operation; this requires an anesthesiologist with experience caring for TCS children.

The craniofacial region is divided into upper and lower sections; the dividing line is at the nose-lip junction. The upper ridge and roof of the *orbits* slants downward (so-called "teardrop" shaped orbits). For the upper face, a surgical procedure can be done to correct this downward slant of the eyes and orbits. The floor of the orbits is deficient or cleft; the zygomas (cheekbones) are small or absent. Bone grafts are used to fill in the clefting and to augment the underdeveloped cheekbones and orbital floors. Usually it is preferable to wait until the child is at least 6-7 years old for sufficient bony thickness in the cranium for harvesting bone grafts. There is a remarkable tendency for bones to dissolve when placed over the cheekbones of children with TCS; therefore, many children require more than one bone graft augmentation.

The two jaws that comprise the lower face are evaluated as the child gets older. The upper jaw (maxilla) is tilted downward and it is small in width and posterior in height. The lower jaw (mandible) is small in its vertical height and tilted downward with an underdeveloped chin. In some children, simple orthodontic treatment in adolescence is all that is necessary to correct the bite. If the chin is retruded, it can be advanced; this procedure (*genioplasty*) is usually postponed until adolescence. In children with more severely expressed TCS, an operative procedure on both the upper and lower jaws is indicated to lengthen the posterior jaw region. For some of these children, lengthening of the lower jaw is done in the 6-8 year age range. Lengthening the mandible can improve the airway. Usually, the final correction of the jaws is undertaken when the child has completed skeletal growth.

There is no cosmetically accepted operation to create lower eyelashes. Reduction of the nose (rhinoplasty) helps to balance the facial features. The "side burns" can be positioned backward in a male or excised in a female- this will leave a scar on each cheek. Depilation (hair removal) is an alternative solution.

Glossary

Autosomal dominant disorder	a condition caused by a mutated gene or several genes that begins spontaneously; thereafter, a 50% chance of appearing in every new child of an affected parent.
Choanae	the funnel-shaped, posterior openings of the nose, just above and behind the hard palate.
Chromosome	strands of genetic material (genes) within the nuclei of cells that determine our structure; each cell contains 46 chromosomes- 23 pairs (one -half from the mother, one-half from the father); the sex chromosomes are one of these pairs.
Coloboma	a slit or fissure in any part of the eye or eyelid.
Depilation	hair removal by a needle and electric current.
Dysostosis	abnormal formation of bone that is biochemically normal.
Expressivity	the degree to which a genetic disorder affects the body structures, usually said to be "variable expressivity", meaning that not all affected individuals have the same severity of an abnormality.
Familial	those behavioral and physical characteristics (traits) determined by gene(s), i.e., the characteristics of a family that are common, more so than in the general population. Familial traits are considered normal or abnormal variations.
Gene	chromosomes are made of genes-nucleic acids (DNA) that program everything of which we are composed, how our body works, and the way we walk, talk, and think
Genetic	refers to an abnormality that results from an alteration in a gene.

Genioplasty	a surgical procedure to alter the position/ appearance of the chin.
Microtia	small, abnormally-shaped, or absent external ear.
Mutation	any permanent, heritable change in the sequence of DNA in a chromosome.
Orbit	the bony socket that contains the eye.
Penetrance	an "all-or-none" phenomenon, referring to whether or not a family member has or does not have a particular genetic abnormality.
Syndrome	several malformations occurring in different areas of the body.
Zygoma	the bony arch from the face to the skull, the "cheek-bone".