

a guide to understanding
**treacher collins
syndrome**

a publication of children's craniofacial association

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this parent's 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.

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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.

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what Is treacher collins syndrome?

Treacher Collins syndrome is a condition in which the cheekbones and jawbone 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 having very small cheekbones and notches in their lower eyelids. This diagnosis is given to children who have notching or stretched lower eyelids and partially absent cheekbones. Their ears are frequently abnormal and part of the outer ear is usually absent. The lower jaw is also small.

why does treacher collins syndrome happen?

There are two ways that Treacher Collins syndrome develops. First, Treacher Collins can develop as a new mutation. This means that both parents pass on normal genes to their child. However, sometimes very early in development a change in one of the genes leads to this mutation. At the present, 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. It is not until a child is born with the syndrome that it is realized that the mother or father also has the condition.

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

It is estimated that Treacher Collins syndrome occurs in one of 10,000 births. For unaffected parents with one child with Treacher Collins, the chance of giving birth to a second child with the condition is negligible. 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 genes, the children may be affected in varying degrees. The degree may be the same as the parent, milder, or more severe.

what problems can be expected?

Several problems are common to Treacher Collins syndrome. A child does not necessarily have all of these problems. The most common difficulties involve breathing, ears, eyes, and hands.

why are there breathing problems?

Children with this condition usually have small underdeveloped jaws. This causes the tongue to be positioned farther back toward the throat, resulting in a smaller airway. The airways become even smaller when these children develop colds and infections because of congestion and swelling.

are these breathing problems a cause for concern?

they can be. One of the most important things to do at an early age is to have your child tested with a sleep study to make sure that he/she is not developing sleep apnea. Sleep apnea is a condition in which the child is not getting enough oxygen while sleeping. The child may even stop breathing for a time. It is now believed that sleep apnea may affect the child's mental development.

what can be done to prevent this situation?

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

is this risk of sleep apnea the only effect of the difficulty with breathing?

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 get enough food to eat. Therefore, they have trouble gaining weight and may require a gastric tube for supplemental feeding. Finally, breathing problems over a long time can eventually affect the heart. Obviously, these conditions require close monitoring by a pediatrician.

what kinds of ear problems are expected?

Most children with Treacher Collins have an abnormality of the external ear. These deformities range from a prominent ear to a complete absence of the external ear.

do these children have hearing loss?

It is important to have the child's hearing tested at an early age. Most children have a 40% hearing loss in each ear. Although it is possible for children to hear without an outside opening in the ear, the sound will be quite muffled. It is very important to get a hearing aid when the child is developing speech in the first few years of life.

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

the sounds that babies hear in the first year are very important for speech development. It was previously believed that most children with Treacher Collins had mental deficiencies. It is now realized that they are as intelligent as other children when given appropriate devices so that they can hear properly in school and other environments.

what are the problems involving the eyes?

the eyes are one of the most noticeable features of Treacher Collins. These children are often described as having a sad appearance. This is caused by the drooping look of the lower eyelids. This appearance can be partially corrected through surgery. It is very important that Treacher Collins patients be followed by an ophthalmologist. Eye ointments may need to be used at night to prevent the eyes from drying out which can lead to infections.

do all treacher collins children have problems with their hands?

avery small percentage of children will have some problem with their hands. One of the more common problems involves the thumbs. The thumbs are either absent or abnormally small. A bilateral absence (absence of both thumbs) may mean that the child has a syndrome called Nager syndrome, which is similar to Treacher Collins syndrome. There are a number of available treatment options that can greatly improve a child's hand function. The treatment used will depend upon the nature of the problem.

when is the best time to correct these problems?

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. It is believed that the best approach is to use the child's own bone and to avoid placing artificial materials beneath the skin. At the time of this surgery, the outer corners of the eyes are usually raised to tighten the lower eyelids.

when can work begin on the ears?

Reconstruction of the ears usually begins at age six. External ears can be built from the child's own rib cartilage. It usually takes three or four operations to finish building the outer ear. Some centers operate on both ears at once to decrease the number of surgeries for the child. After the outer ear is built, some children will be candidates for inner ear surgery to build an ear drum. Other children may be candidates for hearing aid implants. These implanted devices are less noticeable than the traditional hearing aids.

is there surgical correction for the shortened jaw?

Surgery to lengthen the jaw usually occurs in stages. The timing depends on the degree of smallness of the jaw. 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 jaw is usually lengthened by a technique called distraction osteogenesis, which uses a device to gradually lengthen the bone. This technique may decrease the number of surgeries needed to correct the jaw abnormality.

where should I go to seek treatment for my child?

your child should be treated by a qualified craniofacial team. Treacher Collins syndrome is a complex problem. It requires the expert skill of many different specialists working together. These problems are best treated at a craniofacial center.

what is a craniofacial team?

a craniofacial team is a group of specialists specifically trained in the surgical management of problems involving the face and head. The basic team members and their roles in the treatment of your child are detailed in the information that follows.

Anthropologist – This specialist takes multiple measurements of the face and skull. These measurements are compared to tables of normal values. An example of these measurements is the distance between your eyes. Craniofacial surgeons use the information gathered by the anthropologists to see how a child is growing and developing. This also assists in the planning for corrective surgery.

Craniofacial Surgeon – This individual is usually the craniofacial team leader and frequently coordinates the appointments with the other doctors. The craniofacial surgeon will have usually completed general and plastic surgery training, and a fellowship in craniofacial surgery at a children's hospital.

Geneticist – This physician specializes in categorizing different syndromes. This is very important to the overall care of the child. After the geneticist makes the diagnosis, the team members can then look for problems which may be associated with that condition or syndrome and perhaps prevent them. The geneticist also counsels families as to the possibility and probability of future generations having Treacher Collins syndrome.

Neuro-ophthalmologist – This physician will closely follow your child's eyesight and closely monitor any problems. The doctor can perform surgery to balance the eye muscles if there are problems in looking straight ahead with both eyes.

Neuro-radiologist – This physician is specially trained to read x-rays and scans of the brain and the skull. This specialist provides important information to the craniofacial surgeon and neurosurgeon.

Pediatric Anesthesiologist – This doctor is a very important part of any craniofacial team. Children with craniofacial problems often have problems associated with the airways that create breathing difficulties. It is essential that this doctor be well trained in pediatric anesthesiology, but it is just as important that he/she have substantial experience in dealing with these special children. The pediatric anesthesiologist's amount of experience with craniofacial problems perhaps has the greatest effect on the overall safety of the surgery.

Pediatric Dentist – Since children with craniofacial problems often have problems with their teeth, the pediatric dentist will care for these specialized problems. There are circumstances in which teeth are absent or a patient may be unable to open his/her mouth. This makes the care of his/her teeth difficult; therefore, the special skills of a pediatric dentist are needed.

Pediatric Intensivist – This is a pediatrician who specializes in the care of children in intensive care units. This specialist's expertise is called upon to monitor children during the first night following surgery to insure that all goes well.

Pediatric Neurosurgeon – This doctor works with the craniofacial surgeon in the operating room and contributes substantially to the safety of the procedure. This doctor has completed training in neurosurgery and has taken advanced training in pediatric neurosurgery.

Pediatric Nurse – Of all the team members, the pediatric nurse will probably spend the most time with your child. This nurse has specialized training not only in the treatment of children, but specifically the treatment of children with craniofacial conditions.

Pediatric Otolaryngologist – This specialist plays an important role in monitoring the child's hearing. (It has been found that even small improvements in a child's ability to hear can greatly affect his performance in school.)

Pediatric Psychologist – This individual performs two important functions. The first function is to monitor a child’s development to determine a need for intervention in helping your child reach his potential. Second, this individual helps your child to cope with the stress and pressures arising from his/her medical condition. The psychologist can often provide parents with suggestions for dealing with interpersonal relationships. This is especially helpful with handling problems with children at school.

Social Worker – This person often introduces children to the hospital and helps them prepare for surgery. With the rising costs of medical care, the social worker can also help families by providing important financial information.

Speech Therapist – This specialist evaluates your child’s ability to communicate. The trained ear of the speech therapist can sometimes catch early problems that can be corrected with simple speech exercises.

what are the advantages of treatment at a craniofacial center?

Centers with large craniofacial teams working together have the advantage of greater experience. This definitely leads to better results and fewer complications. In addition, ongoing research at these centers offers patients the latest breakthroughs in treatment and technology. As there are only a few experienced centers in the country, it is not uncommon for families to travel long distances to get quality care. By contrast, children treated by individual physicians not working as a team or by inexperienced teams are at a risk of unsatisfactory results. These children sometimes require two or three additional operations to correct procedures performed under these conditions.

are there other advantages of receiving care at a craniofacial center?

another advantage of receiving care at one of the large craniofacial centers is that often more than one operation can be performed by different specialists at the same time. This decreases the total number of surgeries a child will need. Every effort is made to minimize the time a child spends in the hospital. This is important for your child's development, as well as for financial reasons. Having a child with Treacher Collins syndrome can place an enormous financial hardship on the family. It is important to provide surgical correction with the lowest complication rate and with the shortest hospital stay. Craniofacial centers with a qualified staff are equipped to accomplish this with the least amount of physical, emotional, and financial strain.

are there other benefits?

another benefit of traveling to busy, qualified craniofacial centers is the opportunity to meet other children and families affected with similar conditions. These families often share their experiences and offer valuable advice. This provides a tremendous amount of emotional and moral support.

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

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 Web site at www.ccakids.com or call us at 800.535.3643.



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