



**American Cleft Palate-
Craniofacial Association**

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Information about Treacher Collins Syndrome (Mandibulofacial Dysostosis)

What is Treacher Collins Syndrome?

Treacher Collins Syndrome is the name given to a birth defect which may affect the size and shape of the ears, eyelids, cheek bones, and upper and lower jaws. The extent of facial deformity varies from one affected individual to another. A physician named Treacher Collins was one of the first to describe this birth defect. "Syndrome" refers to the group of deformities which characterize affected individuals. Another commonly used medical name for this syndrome is "mandibulofacial dysostosis."

What causes it?

This syndrome is caused by an abnormality in the genes. If both parents are normal, that is showing no signs of the syndrome themselves, this abnormality is the result of a change in the genetic material at the time of conception. The exact cause of this change is not known. If one parent is affected, the abnormal gene is then known to have been contributed by that parent.

Does this mean that this can happen again in my family?

If both parents are normal, the chances of a second child being born with this syndrome are extremely small. However, if one parent is affected, the chances that any pregnancy will result in a child with Treacher Collins is 1 out of 2 (50% risk). For this reason, it is very important that both parents of an affected child be thoroughly examined before any recurrence risks are quoted to them.

If my child, who has Treacher Collins, marries and has children, will all the children have it, too?

No. The risk is 50% for each pregnancy.

What are the risks that my other children will transmit this syndrome to their own children?

If your other children are not affected (showing no signs of the syndrome), there is no increased risk to their children. If another family member shows any feature of the syndrome, the occurrence risk for each pregnancy is 50%.

Will my child be blind or deaf?

There is no evidence that blindness is a feature of this syndrome. Hearing loss, however, is present in most affected individuals to some degree. Early diagnosis and treatment of the hearing loss can prevent associated developmental and educational handicaps.

Will my child be deaf?

The term "deaf" applies only to very severe hearing losses in which the nerves for hearing, in the ear or the brain, do not function properly. The hearing loss in Treacher Collins syndrome is due to abnormalities in the structures of the outer and middle ear which conduct sound to the nerve endings in the inner ear. Thus, the loss in Treacher Collins syndrome is usually termed "conductive" and in the majority of children it is not of sufficient severity to be termed "deafness." However, any degree of hearing loss may affect the development of speech and language ability to succeed in school.

What kinds of problems can I expect?

First, Treacher Collins syndrome, like nearly all birth defects, varies in severity from patient to patient. In fact, some cases are so mild that they are never recognized unless they are seen by specialists experienced in making such a diagnosis. In other children, the physical abnormalities of the face and ears are much more obvious and functional problems may develop.

Second, both the oral cavity (mouth) and the air passage (nose and throat) tend to be small in persons who have this syndrome. This may produce problems for the affected infant with breathing and feeding. You should be on the alert for such problems. If your infant has difficulty breathing or feeding, or has weight loss or poor weight gain, discuss your observations and concerns with your child's primary care provider or craniofacial center. Some children who have severe breathing difficulties require an operation to improve breathing and/or feeding.

Third, cleft palate is a condition frequently associated with this syndrome. Cleft palate itself sometimes can cause feeding problems and increase the risk of middle ear problems. Your child's primary care provider or cleft palate or craniofacial center can assist you with the management of feeding problems.

The next concern after breathing and feeding is hearing. The hearing loss in Treacher Collins syndrome is usually bilateral (meaning that both ears are affected) and, while it is not severe enough to be termed "deafness," it is severe enough to affect the ability to hear the human voice. Hearing levels can and should be measured. Depending upon the results of the testing, your child may be fitted with a hearing aid to restore his or her access to the world of sound. An early childhood program of speech and language therapy may also be recommended.

The fact that a hearing loss is present does not mean that your child will be dependent upon sign language. The great majority of children with this syndrome do learn to talk. However, there are several features of the syndrome, besides the hearing loss, which can affect speech and language development. Particularly in the severely affected child, the size and position of various structures inside the mouth (e.g. the relationship of the upper and lower teeth) may affect the ability to learn certain speech sounds.

You can facilitate your child's speech and language development by (1) seeking early evaluation by a specialist in hearing (an audiologist) and a specialist in communication development (a speech/language pathologist), and (2) follow their advice with regard to the need for a hearing aid and for early therapy programs. The specialists most prepared to evaluate and manage your child are those who are members of a multidisciplinary craniofacial team.

What about other areas of development: social, educational, etc.?

The facial deformity and need for treatment of Treacher Collins syndrome may create problems in family and social relationships, in school adjustment, and so on. The craniofacial center may have a psychologist or social worker, or the center can refer you to someone for evaluation and counseling if needed. Remember that

children with Treacher Collins syndrome, like all other children, are individuals. They vary in social adjustment, academic achievement, and their ability to cope with adults. The professionals of craniofacial centers try to maximize each child's potential by offering early diagnosis and treatment when indicated.

What kind of treatment is available for my child?

First, as explained above, your child may need a hearing aid and this can be determined in the first few months of life.

Second, an early childhood program for speech and language stimulation may be recommended.

Third, if a cleft palate is present, the craniofacial team will advise you on the optimum timing for surgical closure of the cleft.

Fourth, reconstructive surgery is available to improve the appearance of the face. The craniofacial center will advise you on what to expect from such surgery and on optimum timing. Since not all children are affected to the same degree, both the necessity and the outcome of reconstructive surgery vary from child to child.

What should I be doing for my child now?

Be certain that the diagnosis is correct. Treacher Collins syndrome shares some features with other syndromes, and not all physicians are aware of this. For this reason, you are best advised to locate a craniofacial center where genetic consultation, evaluation, and treatment planning will be provided by an experienced multidisciplinary staff composed of representatives from a variety of medical, dental, and other health care specialties. You may not have such a center near your home, but the care your child will receive will be more than worth the inconvenience of traveling to another city. Finally, meet other individuals and families affected by similar facial differences by joining a parent-patient support group.

Please contact ACPA Family Services for further information or for a referral to a cleft palate/craniofacial team.