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# Treacher Collins Syndrome

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Treacher Collins syndrome, also called mandibulofacial dysostosis, is a condition that affects the development of bones and tissues of the head and face. In 1900, it was named after Dr. Treacher Collins, a British ophthalmologist, who described two children as having notches in their lower eyelids and very small cheekbones.

Treacher Collins syndrome is estimated to occur in one of 50,000 births. There are two ways that it can develop. The first way is through a mutation of normal genes from parents. This mutation occurs in very early stages of development, but there is no evidence it is a result of the mother's actions or activities during her pregnancy. The second way it can develop is by inheriting the gene from a parent; this occurs in approximately 40% of cases. It is possible that a parent may have a very mild form of the syndrome that is not detected until the child is born with it.

There are several defining characteristics of Treacher Collins syndrome. These may include:

- downward-slanting eyes, often described as having a "sad" appearance;
- notched lower eyelids;
- underdeveloped or absent cheekbones;
- an underdeveloped or absent side wall and floor of eye socket;
- a small and slanting lower jaw; and/or
- underdeveloped, malformed, and/or prominent ears.

Depending on the severity of many of these physical characteristics, surgery may be required to correct or repair the bones and tissues of the face.

Children with Treacher Collins syndrome may have some difficulties as a result of the development of facial bones and tissues. These may include:

- breathing and/or eating difficulties, due to an underdeveloped jaw;
- 40% hearing loss in each ear, due to abnormalities of the outer and middle ear;
- eye infections, due to a tendency for the eyes to dry out;
- cleft palate; and/or
- absent or abnormally small thumbs.

Children with this syndrome typically have normal development and intelligence; however, early intervention is beneficial for any speech, language, hearing, or feeding difficulties that may arise.

Children with Treacher Collins syndrome should be treated by a qualified craniofacial team. It is a complex syndrome that requires the expertise of many different specialists that work together. Members of the craniofacial team may include a craniofacial surgeon, geneticist, ophthalmologist, radiologist, anesthesiologist, dentist, nurse, otolaryngologist (ear nose and throat doctor), psychologist, social worker, and/or speech-language pathologist.

There are many families and organizations that create a strong support system for those diagnosed with Treacher Collins syndrome and their family members. There are also many books, websites, and videos to supplement these groups. Children with this syndrome often grow and develop into successful adults with successful lives. If you have questions or concerns, please contact a speech-language pathologist or a member of a craniofacial team.

"Treacher Collins Syndrome," Children's Craniofacial Association, accessed June 25, 2018, https://ccakids.org/treacher-collins-syndrome.html.

"Treacher Collins Syndrome," FACES, accessed June 25, 2018, http://www.faces-cranio.org/Disord/Treacher.htm