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Fact Sheet

Treacher Collins Syndrome

Information retrieved from: http://ghr.nlm.nih.gov/condition/treacher-collins-syndrome; http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002624/; http://www.ccakids.com/Syndrome/TreacherCollins.pdf

What is Treacher Collins Syndrome? Treacher Collins syndrome (TCS), also known as mandibulofacial dysostosis, is primarily a hereditary condition that causes underdevelopment of the face, including the eyes and ears. The disorder is named after British ophthalmologist Dr. Edward Treacher Collins, who first diagnosed it in 1900. The signs and symptoms of TCS range from almost unnoticeable to severe and vary in degrees from generation to generation and person-to-person. Some people born with TCS also have cleft palate (an opening in the roof of the mouth). In severe cases, this underdevelopment can restrict the airway, causing potentially life-threatening respiratory problems. Individuals with Treacher Collins syndrome usually have normal intelligence.

What Causes TCS? TCS is essentially a breakdown of the gene protein treacle which causes certain gene mutations. There are a couple of ways in which this can occur. First, TCS can develop as a new mutation. In this case, both parents pass on unaffected genes to their child, but early in the child's development a mutation of the gene occurs. In the second scenario, the child inherits TCS from one of the parents. Inherited cases are often not noticed until the child is born because the parent had such a mild form of TCS that it went undetected.

What is the Prevalence of TCS? Adults with TCS have a 50 percent chance of passing it on in varying degrees of severity to their children. TCS has been estimated to occur in 1 out of every 10,000 births. Parents who are unaffected by TCS may have a child with the disorder. The probability of the parents having a second child having the condition is extremely small.

What are the Signs and Symptoms of Treacher Collins syndrome? Children with TCS may have some, all, or varying degrees of the following: scalp hair that extends to the cheek bones, a defect in the eye extending to the lids, eyes that slant downward, sparse eyelashes, a very small jaw, large mouth, absent, small or unusually formed ears, and/or hand problems. Some affected individuals have additional eye and ear abnormalities that can lead to vision and hearing loss. The latter finding is why all children with TCS should have quality vision and hearing evaluations.

The degree of severity of vision loss occurs because of the presence of disorders such as glaucoma,

strabismus (poor eye alignment), microphthalmia (very small eyes), or coloboma, which is a hole in one of the structures in the eye such as the iris, choroid, retina, and/or optic disc). A coloboma may occur in one or both eyes; its effects on the severity of vision loss depend on the size and location of the coloboma.

Hearing loss occurs in about half of all affected individuals and is caused by defects of the three small bones in the middle ear, which transmit sound, or by underdevelopment of the ear canal. Most individuals experience an average of 40% loss of hearing in each ear. It is possible for children to hear without an external opening but the sound is muted.

How is TCS diagnosed? TCS is most often noticed at birth. In many instances, the child may have difficulty breathing and require a tracheotomy. It is important to seek medical testing and/or treatment as soon as possible. The best resource for diagnosis and treatment is a craniofacial center, where several physicians work as a team to treat and manage problems involving the face and head. This team of physicians may include a craniofacial surgeon, an anthropologist, a geneticist, an ophthalmologist, an audiologist, a radiologist, and a pediatric dentist.

What is the Treatment for TCS? It is imperative that the child or infant get treatment for a coloboma and hearing loss. Eye ointments may need to be used at night to prevent infections; hearing aids enable better hearing and proper speech and language development. Several types of surgeries may be required to reconstruct the ears, rebuild the cheekbones, correct cleft palate, and enable better breathing and swallowing. The timing of the different surgeries varies according to age and severity of TCS. In addition the child may be eligible for special education services that include teachers of students with visual impairments and/or hearing impairments and appropriate related service providers.

Where can People get more information about TCS?

Treacher Collins Foundation:
www.treachercollinsfnd.org

Reflections on Treacher Collins syndrome:
http://www.treachercollins.org/tcs/Welcome.html

The National Craniofacial Association:
http://www.faces-cranio.org/Disord/Treacher.htm

For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project contact:

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Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 4/13