

An autosomal dominant condition, Treacher Collins Syndrome (TCS) was first described in 1900 by Edward Treacher Collins. Predominant traits include facial abnormalities such as a small jaw and low set ears (see Figure 1). TCS is diagnosed in 1 out of 50,000 births annually. Severity of TCS varies from person to person, and from generation to generation.

Characterized by:

Abnormal, low-set, or absent pinnae, micrognathia (very small lower jaw), coloboma (missing pieces of tissue in structures that form the eye), cleft palate, flat cheekbones, and conductive hearing loss.

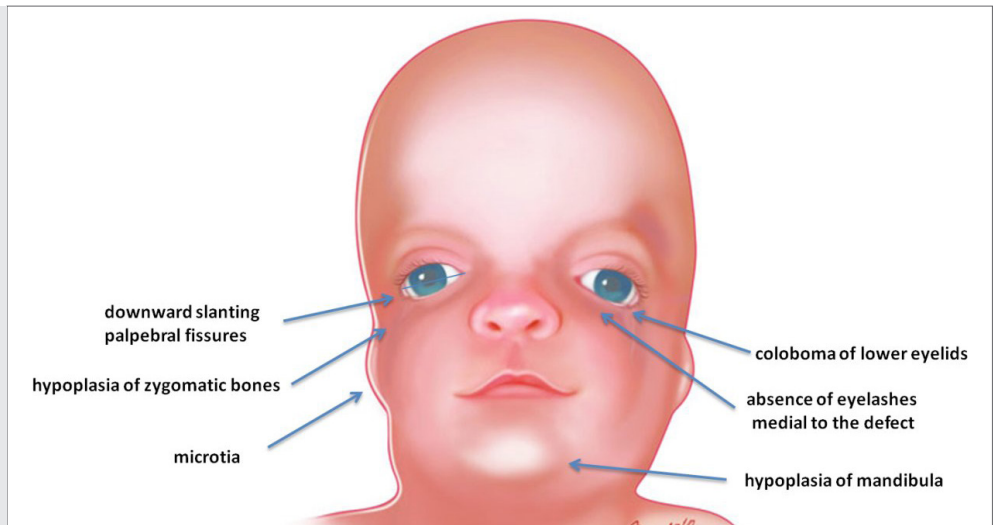


Figure 1. Representation of characteristics seen with Treacher Collins Syndrome. Note low-set pinnae and micrognathia, flat cheekbones, and drooping lower eyelids. From http://www.cranirare.eu/phenotypes/treacher_collins/index.php

Prognosis:

Good. Children diagnosed with TCS typically have normal intelligence and become typically functioning adults. Some may require plastic surgery (e.g., due to cleft palate or micrognathia to aid in feeding); however, surgeries to correct other facial structures may be desired.

Differential Diagnosis:

Usually done by visual exam, but diagnostic tests include X-ray and/or CT scans to visualize internal tissues, bones and organs. Goldenhar Syndrome, Nager Syndrome, Miller Syndrome, Hemifacial Microsomia are other conditions that have similar signs and traits as TCS.

Audiological Findings and Treatment:

Conductive hearing loss can range in severity from mild to severe. In most cases of TCS, malformation of the ossicles is the cause of the conductive hearing loss which can be diagnosed by MRI. The cochlea and auditory nerve are usually not affected. Use of a bone conduction hearing aid is recommended to stimulate the cochlea for hearing during childhood; however, a bone-anchored processor on a softband or a bone-anchored hearing system may be possible if there is good bone thickness in the skull and the candidate is 5 years of age or older. Craniofacial abnormalities may warrant speech and language therapy, in addition to surgery for cleft palate.

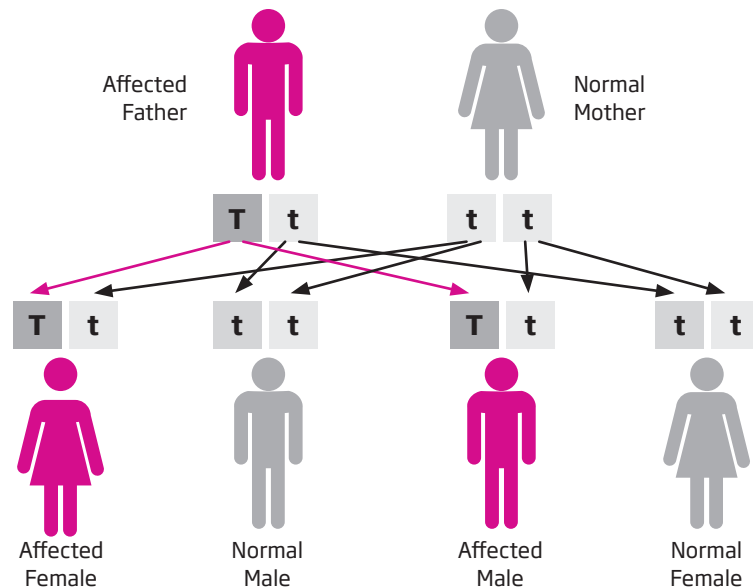


Figure 2. In this example, the likelihood of TCS being passed along to this couple's progeny is 50% regardless of gender. According to literature, 40% of mutations are inherited and 60% of mutations are new. From: <http://www.treachercollins.co.uk/gene/genes.htm>

Gene Expression:

The cause of TCS is unknown, but it is expressed as a mutation of the TCOF1 gene, on chromosome 5, in which haploinsufficiency* of the Treacle protein occurs in the gene. This gene mutation is seen in approximately 95% of those diagnosed with TCS. (See Figure 2)

**Haploinsufficiency occurs when a diploid organism (e.g., human or other mammal) has only one functional copy of a gene. This happens when the other copy of the gene has a mutation and therefore inactivates. The result is the inability of the first copy of the gene to provide the expected function. In the case of TCS, this causes a reduction of neural crest cells during gestational development, leading to a reduced migration of crest cells to the 1st and 2nd pharyngeal arches, which are important for craniofacial development.*

Educational and other Psychosocial Considerations:

Hearing and speech difficulties may lead to educational difficulties. Use of FM along with amplification and preferential seating can provide the child an improvement in SNR while in complex listening environments. TCS should be noted in the child's IEP, as hearing and visual impairments can impact their performance in classroom abilities. Affected children and their families may need a great deal of support. Integration into social activities should be discussed and practiced to encourage immersion in groups and to discourage bullying from others.

Professional Considerations:

Support team may be large and can include the child's pediatrician, oral/maxillofacial surgeon, ENT, child social worker or psychologist, speech pathologist, audiologist, and developmental specialist.

Online Support and References:

<http://ghr.nlm.nih.gov/condition/treacher-collins-syndrome>
<http://www.primehealthchannel.com/treacher-collins-syndrome.html>
<http://ghr.nlm.nih.gov/gene/TCOF1>
<http://www.treachercollins.co.uk/gene/genes.htm>
<http://www.treachercollins.net/>
<http://www.asha.org/Research/reports/craniofacial/>
<http://health.dir.groups.yahoo.com/group/treacher/?v=1&t=directory&ch=web&pub=groups&sec=dir&slk=1>
<http://0-www.nlm.nih.gov.sapl.sat.lib.tx.us/medlineplus/ency/article/001659.htm>

Other References:

Marres, H. A. (2002). Hearing loss in the Treacher-Collins syndrome. *Advances in Oto-rhino-laryngology*, 61, 209-215.
 Marszarlek, B., Wojcicki, P., Kobus, K., & Trzeciak, W. H. (2002). Clinical features, treatment, and genetic background of Treacher Collins syndrome. *Journal of Applied Genetics*, 43(2), 223-233.
 Splendore A, et al. (2003) Parental origin of mutations in sporadic cases of Treacher Collins syndrome. *Eur J Hum Gen*, 11, p. 718-722