Treacher Collins Syndrome/Mandibulofacial Dysostosis

What is Treacher Collins Syndrome?

Treacher Collins Syndrome, also called mandibulofacial dysostosis, affects the head and face. Characteristics include:

- Down-slanting eyes
- Notched lower eyelids
- Underdevelopment or absence of cheekbones and the side wall and floor of the eye socket
- Lower jaw is often small and slanting
- Forward fair in the sideburn area
- Underdeveloped, malformed and/or prominent ears
- Most children with Treacher Collins have normal development and intelligence; however, it is important that there be early hearing tests. Most children with Treacher Collins Syndrome benefit from early intervention speech and language programs.

What are the signs and symptoms of Treacher Collins syndrome?

The signs and symptoms of this disorder vary greatly, ranging from almost unnoticeable to severe. Most individuals have:

- underdeveloped facial bones,
- particularly the cheek bones, and
- A very small jaw and chin (micrognathia).

Some people with this condition are also born with an opening in the roof of the mouth called a cleft palate. In severe cases, underdevelopment of the facial bones may restrict an affected infant's airway, causing potentially life-threatening respiratory problems.

Causes

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**." This syndrome is **caused** by an abnormality in the genes.

Diagnosis

Treacher Collins syndrome (TCS) is caused by changes (mutations) in any of several genes: *TCOF1* (in over 80% of cases), *POLR1C*, or *POLR1D*. In a few cases, the genetic cause of the condition is unknown.

These genes appear to play important roles in the early development of bones and other tissues of the face. They are involved in making proteins that help make ribosomal RNA (rRNA). rRNA is a chemical needed to make new proteins that are necessary for normal function and survival of cells. Mutations in these genes can reduce the production of rRNA, which may cause cells involved in the development of facial bones and tissues to die early. This premature cell death may lead to the signs and symptoms of TCS. It is still unclear why the effects of these mutations are generally limited to facial development.[1]

Treatment

There is currently no cure for Treacher Collins syndrome (TCS).[4] Treatment is tailored to the specific needs of each affected person. Ideally, treatment is managed by a multidisciplinary team of craniofacial specialists.

Newborns may need special positioning or tracheostomy to manage the airway. Hearing loss may be treated with bone conduction amplification, speech therapy, and/or educational intervention.[5]

In many cases, craniofacial reconstruction is needed. Surgery may be performed to repair cleft palate, to reconstruct the jaw, or to repair other bones in the skull. The specific surgical procedures used and the age when surgery is performed depends on the severity of the abnormalities, overall health and personal preference.[4]

Resources

- **MedicineNet.com: Treacher Collins Syndrome -** www.medicinenet.com/treacher_collins_syndrome/article.htm
- **Genetic and Rare Diseases Information Center -** https://rarediseases.info.nih.gov/diseases/9124/treacher-collins-syndrome
- **Wikipedia -** https://en.wikipedia.org/wiki/Treacher_Collins_syndrome
- **FACES: The National Craniofacial Association -** www.faces-cranio.org
- **WebMD -** www.webmd.com/children/treacher-collins-syndrome-10671