

Treacher–Collins syndrome

Also known as: mandibulofacial dysostosis, Franceschetti syndrome

Clinical Characteristics

Treacher-Collins syndrome is characterized by underdevelopment of the midface. This includes a small or receded jaw and chin, notching of the lower eyelid, sparse or absent eyelashes, a prominent nose, cleft palate with or without cleft lip, hair displacement in which hair grows from in front of the ear towards the cheekbones, external ear abnormalities such as microtia or anotia, rotated ears, and conductive hearing loss. Individuals with Treacher-Collins syndrome are sometimes described as having a “sunken” or “fishlike” appearance to their midface.

Condition and Hearing Loss

40-50% of patients with Treacher-Collins syndrome have conductive hearing loss, most often due to malformations of the ossicles. The middle ear cavities may also be underdeveloped. The structures of the inner ear tend to be normal. Hearing loss is usually bilateral.

Natural History

Treacher-Collins syndrome is diagnosed in both males and females, and in all ethnicities. The prevalence of the syndrome is estimated to be between 1 in 10,000 and 1 in 70,000.

There is wide clinical variability to the syndrome, even among members of the same family. Some people are so mildly affected that they go undiagnosed, while others have life-threatening manifestations. The features of Treacher-Collins syndrome are classically bilateral and apparent at birth. Some infants may require intervention if breathing or feeding is obstructed. The most common clinical features are midface hypoplasia (found in around 90%), micrognathia/retrognathia (78%), external ear abnormalities (77%), and notching of the lower eyelid (69%). With proper intervention for any early breathing or feeding problems, the health of individuals with Treacher-Collins syndrome is good, overall. Intelligence is usually normal. Fertility is normal. Life expectancy is normal.

Genetics

The gene *TCOF1* on the long arm of chromosome 5 is the only known gene associated with Treacher-Collins syndrome. The normal gene product, treacle protein, appears to be associated with the ribosomes of the cell, though the contribution of this to the characteristics of the syndrome is not well-understood. Approximately 60% of

individuals with Treacher-Collins syndrome have a new mutation and thus there is no history of the disorder in the family. The other 40% inherited a mutation from an affected parent. Treacher-Collins syndrome is **autosomal dominant**. This means that an affected individual has a 50% chance with each pregnancy of having an affected child, and a 50% chance of having an unaffected child. Since Treacher-Collins syndrome is so variable in its appearance, seemingly unaffected parents of an affected individual should undergo clinical examination to ensure that neither one is mildly affected and has missed diagnostic attention previously.

Management

The diagnosis of Treacher-Collins syndrome is made by clinical evaluation and radiographic findings. Genetic testing is clinically available for confirmation of diagnosis, and prenatal diagnosis if a mutation has been identified in an affected family member.

Infants with Treacher-Collins syndrome should have their airway assessed, as well as feeding and breathing abilities. A craniofacial CT scan should be performed in the first six months of life to document head and neck structures. Formal audiologic examination and ophthalmologic evaluation should also be done.

Management should be handled by a multidisciplinary craniofacial team, including a medical geneticist, plastic surgeon, head & neck surgeon, otolaryngologist, oral surgeon, orthodontist, audiologist, speech pathologist, and psychologist. In the early years, proper positioning of the airway and a gastrostomy tube could be needed. Craniofacial reconstruction and orthodontic surgery will need to be scheduled carefully over the course of several years to prevent the progression of facial asymmetry. Bone reconstruction should precede surgeries involving the soft tissues.

Hearing loss is often treated by means of bone conduction amplification, speech therapy, and varied educational interventions. Individuals with ear anomalies may require a bone-anchored hearing aid. External ear reconstruction is included in a long-term surgical plan. The plan may also include external auditory canal and middle ear reconstruction for those with microtia. Regular audiologic screening is recommended.

Many individuals with Treacher-Collins syndrome face social challenges as they grow older. Children and adolescents may face teasing or rejection due to their appearance. These issues must be handled sensitively and openly. Many people find that talking to a counselor or psychologist helpful, while others find their own coping strategies. This should be discussed with parents at length while their child is still young so they may be prepared for such issues in the future.

Resources for Families

Statewide Genetics Program

Phone: 608-267-7148

Fax: 608-267-3824

Email: meyeram@dhfs.state.wi.us

Wisconsin First Step Hotline

Phone: 1-800-642-7837 voice/TTY

Website: www.mch-hotlines.org

Wisconsin Office for Deaf and Hard of Hearing

Phone: 1-608-266-3118 voice/TTY

Website: www.dhfs.state.wi.us/sensory

Regional Children and Youth with Special Health Care Needs Centers

Centers in Green Bay, Wausau, Milwaukee, Madison, and Chippewa Falls

Website: http://dfhs.wisconsin.gov/DPH_BFCH/cshcn/index.HTM

WI Chapter of Families for Hands & Voices

Phone: (920) 437-7370

Website: www.handsandvoices.org

Parent-to-Parent of Wisconsin

Phone: 1-888-266-0028

Email: rmathea@shsmh.org

Family Village online resource

Library Card Catalog of Disorders

www.familyvillage.wisc.edu

FACES: The National Craniofacial Association

P.O. Box 11082

Chattanooga, TN 37401

Phone: 1-800-332-2373

Email: faces@faces-cranio.org

Website: www.faces-cranio.org/Disord/Treacher.htm

National Organization for Rare Disorders (NORD)

www.rarediseases.org