Treacher Collins Syndrome (TCS) - A Review

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Abstract:

Treacher Collins syndrome is predominantly a complex orofacial disorder. This syndrome without a management can lead to a life threatening condition. It has a very characteristic facial appearance so that it can be easily recognized. Typical features comprise antimongoloid palpebral fissures, coloboma of the eyelid, hypoplasia of the malar bones, hypoplasia of the mandible. So this review illustrates the etiology, clinical features, differential diagnosis and management of Treacher Collins syndrome.

Keywords:

Treacher Collins syndrome, Orofacial disorder, Cleft palate

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Introduction:

Thomson in 1846 described this syndrome, but it was E. Treacher Collins who expressed its essential components in 1900. In 1949, Franceschetti and Klein carried out extensive studies. Other suggested names include mandibulofacial dysostosis, Berry's syndrome and Franceschetti-Zwahlen-Klein syndrome. The estimated incidence of Treacher Collins syndrome ranges from

1:40,000 to 1:70,000 of live births.[1] This review portrays the etiology, clinical features, differential diagnosis and management of Treacher Collins syndrome

Etiology:

Mutation in the *TCOF1 gene*, at chromosome 5q32-q33 and treacle the protein coded by this gene has been hypothesized to assist in protein sorting during particular stages in embryonic development. Recently investigators are pondering the teratogenic role of hypervitaminosis-A.[2]

Clinical forms:

Franceschetti and Klein described five clinical forms[3]

(1) The complete form (known features)

(2) The incomplete form (presenting variably with less severe ear, eye, zygoma, and mandibular abnormalities)

(3) The abortive form (only the lower lid pseudocoloboma and zygoma hypoplasia are present)

(4) The unilateral form, (anomalies limited to one side of the face)

(5) The atypical form (combined with other abnormalities not usually part of the typical syndrome).

Clinical features:

Major clinical features[4]

• *Hypoplasia of the zygomatic bones and mandible* resulting in the following:

– Midface hypoplasia (89%)

– Micrognathia and retrognathia (78%)

• *External ear abnormalities* (77%) including absent, small, and malformed ears (microtia) or rotated ears

• *Lower eyelid abnormalities* including the following:

– Coloboma (notching) (69%)

– Sparse, partially absent, or totally absent cilia (lashes) (53%)

• *Family history* consistent with autosomal dominant inheritance (40%)

Minor clinical features[4]

• *External ear abnormalities* including atresia or stenosis of the external auditory canals (36%)

• *Conductive hearing loss* (40%-50%)

• *Ophthalmalogic defects*

– Vision loss (37%)

– Amblyopia (33%)

– Refractive errors (58%)

– Anisometropia (17%)

– Strabismus (37%)

• *Cleft palate* with or without cleft lip (28%)

• *Preauricular hair displacement* (26%), in which hair growth extends in front of the ear to the lateral cheekbones

• *Airway abnormalities* including the following:

– Tracheostoma

– Uni- or bilateral choanal stenosis or atresia

• *Delayed motor or speech development*

• *Hypoplasia or aplasia (discontinuity) of the zygomatic arch*

• *Malar hypoplasia*

• *Mandibular retrognathia*

1. Skull is Square forehead, flattening of the occipito-parietal region, lack of angulation in the naso-frontal region and long occipito-frontal diameter, asymmetry of the skull.[5]

2. Facial abnormalities are bilateral and usually symmetric, but not always. The large appearance of the nose is secondary to hypoplastic supraorbital rims and hypoplastic zygomas. Downward-sloping palpebral fissures, depressed cheekbones, malformed pinnae, receding chin, and large down-turned mouth are characteristic.[6]

3. Blind fistule on the cheeks, and occasionally characteristic "tongue-shaped" growth of hair on the lateral side of the face.[5]

4. Middle ear abnormalities include irregular or absent auditory ossicles with fusion between rudiments of the malleus and incus, partial absence of the stapes and oval window. Consequently, bilateral conductive hearing loss is common in TCS patients.[7]
5. Oral manifestations reported are palatal cleft, congenital palatopharyngeal incompetence, rarely cleft lip-palate, macrostomia (unilateral or bilateral), elevator muscles of the upper lip are deficient, parotid salivary glands may be absent or hypoplastic, pharyngeal hypoplasia (frequent finding for neonatal death).[6]
6. Atresia of pharyngeal ring.[5]
7. Big nose with a very short columella, narrow nares and small lateral cartilages.[5]

Differential diagnosis:

a) *1st branchial arch syndrome:* Macrosthomia, hemignathia and tragus abnormalities.[8]

b) Other anomalies, such as the Crouzon Disease, Apert Syndrome, hyperterolism, Möbius Syndrome and Romberg disease.[8]

Management:

A multispecialist approach with the participation of pediatricians, ENT, and speech therapists, with the significant role being the surgical correction. The most severe forms of the disease require lifesaving surgery at early infancy. Retrusion and shortening of the mandible, as well as posterior position of the tongue, result in obturative insufficiency of the upper airways. In children pharyngeal diameter smaller than 1 cm can develop nocturnal apnea leading to sudden death. Such patients require tracheostomy and sometimes even gastrostomy in the first few days after birth.[9]

Patient with TCS syndrome may need a hearing aid at an early age depending on severity of hearing loss. The sounds children recognize in first year of development are important for speech development. Children who are treated at an early age with hearing aids were as intelligent as other children.[10]

Conclusion:

TCS is a complex life threatening syndrome requires an early management for survival. Early treatment for auditory abnormalities are must to have a normal intelligence. Psychological counseling for those patients with cosmetic abnormalities is required. So role of Dental specialties play a major role in this syndrome.

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