Treacher Collins Syndrome (TCS) and its Oromaxillofacial Features: A Review

Dr. Md. Asad Iqubal¹, Dr. Naushad Anwar², Dr. Mobeen Khan³, Dr. Chandra Prakash Gupta⁴, Dr. Nazish Akhtar⁵, Dr. Divyanshu Shrivastava⁶.

Senior Lecturer, Department of Oral Medicine and Radiology, Patna Dental College and Hospital, Patna, Bihar, India¹.
Tutor, Department of Oral Medicine and Radiology, Patna Dental College and Hospital, Patna, Bihar, India².
Senior lecturer, Department of Oral Medicine and Radiology, Chandra Dental College and Hospital Barabanki, Uttar Pradesh, India³.
MDS, Department of Orthodontics, Chandra Dental College and Hospital Barabanki, Uttar Pradesh, India⁴.
Senior Lecturer, Department of Oral Medicine and Radiology, Seema Dental College and Hospital, Rishikesh, India⁵.
Post Graduate Student, Department of Pedodontics and Preventive Dentistry, Chandra Dental College and Hospital Barabanki, Uttar Pradesh, India⁶.

Corresponding Author: Dr. Mobeen Khan Email ID: drmkhan26@gmail.com

Abstract:

Treacher Collins Syndrome (TCS) is a rare autosomal dominant disorder of craniofacial development. Major features of Treacher Collins Syndrome are midface hypoplasia, micrognathia, microtia, conductive hearing loss, and cleft palate. It is named after E Treacher Collins who described the essential components of the condition in 1900. Incidence of this syndrome is approximately 1 in 40,000-70,000 live births and it affects both genders equally. It affects structures which are derivatives of first and second branchial arches. The most common manifestations of Treacher Collins Syndrome are the antimongloid slanting of palpebral fissures, colobomas of lower eyelid, hypoplasia of zygoma and mandible; and a variety of ear abnormalities.
Introduction:

Treacher Collins Syndrome - or mandibulofacial dysostosis – is a rare condition. It presents several craniofacial deformities of different levels. This is a congenital malformation involving the first and second branchial arches. The disorder is characterized by abnormalities of the auricular pinna, hypoplasia of facial bones, antimongoloid slanting palpebral fissures with coloboma of the lower eyelids and cleft palate. Treacher Collins Syndrome is rarely associated with choanal atresia.\(^1\)

Several hypotheses were proposed to explain the pathogenesis of TCS including abnormal patterns of neural crest cell migration, abnormal domains of cell death, improper cellular differentiation during development or an abnormality of the extracellular matrix. Mann and Kilner assumed the etiology to be an inhibitory process occurring towards the seventh week of the embryonic life and affecting the facial bones deriving from the first visceral arch. John Mckenzie suggested that the cause of the abnormality is a defect of the stapedial artery which causes mal-development in its own field as well in the region of first visceral arch.\(^2\)

Oromaxillofacial Features: Following are the common features:

- Microtia with possible hearing loss.
- Hypoplastic cheeks, zygomatic arches and mandible.
- Anti mongoloid slant to the eyes.
- High arched or cleft palate.
- Malocclusion (anterior open bite).
- Macrostomia (abnormally large mouth).
- Colobomas.
- Pointed nasal prominence.
- Increased anterior facial height.
- Small oral cavity & airway with normal sized tongue.

Fig: 1. Front View of patient showing asymmetrical face, depressed cheek bones, relatively large nose, micrognathia, scanty lower eye lashes, downward sloping palpebral fissures.

Fig: 2. Profile view of patient showing bird face, retruded chin and malformed ear.
The most frequent clinical manifestations, among a great variety of alterations, include: antimongoloid slanting of palpebral fissures (89%), malar hypoplasia (81%), mandibular hypoplasia (78%), auricular pinna malformations (77%) and lower palpebral coloboma (69%).

TCS is a well-recognized condition characterized by variable involvement of the craniofacial structures derived from the first and second branchial arches. Clinically, it ranges from hypoplasia of the zygomatic arches and antimongoloid slant of the palpebral fissures, which are considered the minimal diagnostic features, to a more complex phenotype with skeletal, cardiac, and renal manifestations.
Teber et al. (2004)\(^6\) defined the downward slanting palpebral fissures and the hypoplasia of the zygomatic arches as the minimal features for the diagnosis of TCS. Early diagnosis of TCS allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients. If this can be done early, it is possible to take advantage of anticipated growth during normal skeletal maturation and to obtain better therapeutic results.\(^7\)

The craniofacial skeleton presents abnormalities in the mandible, maxilla, zygomatic areas, orbits, ears and skull base. The height of the mandibular branch is deficient and the length of the mandible body is reduced. The distortion which is present in the symphysis menti contributes for the mandibular deficiency and for the increase of the inferior height of the face, which leads to an increase of the cranial-base-mandibular angle. The mentum angle may also be bigger than the standard. Consequently, the mandible in TCS carriers is retrognatic, the temporal-mandibular articulation is anteriorly misplaced, the mandibular angle is obtuse and the mandible is smaller than the maxilla.\(^8\)

**Clinical Forms of TCS:**

Franceschetti and Klein\(^9\) described five clinical forms:

1. The complete form (having all 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 face) and
5. The atypical form (combined with other abnormalities not usually part of typical syndrome).

**Medical Management of Treacher Collins Syndrome:**

A multidisciplinary team is required for the medical management of patients suffering from Treacher Collins Syndrome. The multidisciplinary team must be consists of following department of health sciences:

- Craniofacial surgeon
- Ophthalmologist
- Speech therapist
- Oral and Maxillofacial health care unit
- ENT surgeon
- Otorhinolaryngologist

**Conclusion:**

Treacher Collins Syndrome must continue being investigated, so that it is possible to improve the current approach in functional and aesthetical corrections. The management of TCS must be done by multidisciplinary approach. Early diagnosis can improve the quality of life.

**References:**


