Treacher Collins Syndrome the Pivotal Role of its Prompt Diagnosis- A Case report

Abstract:
Mandibulofacial dysostosis, also called Franceschetti-Zwahlen-Klein syndrome or Treacher Collins syndrome (TCS), is identified by these terms. Atypical differentiation of the first and second pharyngeal arches during fetal development is linked to Treacher Collins syndrome (TCS). This syndrome is equally prevalent in both genders and affects about 1 in 50,000 live births. Facial bone hypoplasia may be the first sign of the disorder. We present a case report of Treacher Collins syndrome with their extra oral findings, intraoral findings and their treatment plan. Furthermore, we encompassed a range of etiological elements, clinical diagnostic devices, and multidisciplinary teamwork.

Key-words: Treacher Collins syndrome (TCS), Franceschetti-Zwahlen-Klein syndrome, Mandibulofacial dysostosis, Clinical features

Introduction:

Treacher Collins syndrome (TCS) is an autosomal dominant disorder that affects the development of structures originating from the first and second brachial arches during early embryonic development.[1] The essential components of the syndrome were described by Edward Treacher Collins (1900), the ophthalmologist after whom the disease is named.[2] Treacher Collins syndrome (Mandibulofacial dysostosis) is characterized by deafness, facial bone hypoplasia, chin and cheekbones), anti-Mongoloid tendency to clefts, lower eyelid coloboma and bilateral ear disorders.[3] The quality of life of these people can be improved by early detection and treatment.[4]

Case Report:

A 7-year-old boy reported to the department of Oral medicine & Radiology, Sardar Patel Post Graduate Institute of Dental & Medical Sciences Lucknow; complaining of deformity of face since birth. The patient's father also complained of asymmetry of his face, small size lower jaw and deafness (Fig. 1).

On general examination, the patient was 3 feet 8 inches tall and had small body frame was observed. He weighs 21 kg, moderately nourished and had a normal posture & gait.

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Extraoral Findings:

Ophthalmological findings: Downward slant of corner of eyes, Tear drop shaped orbits, Hypertelorism, Absence of lashes along medial one-third of lower eyelid

ENT findings: Slightly broadened nasal bridge (Fig. 2)

Facial dysmorphic features: Symmetrically disfigured face, Hypoplasia of malar bone, Hypoplasia of lateral aspect of orbit, Hypoplastic mandible/ underdeveloped chin, tongue shaped extension of temporal hair onto the cheek(Fig. 3)

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Intraoral Findings :

Malocclusion, crowding in lower anterior, High arched palate, mouth breathing, V-shaped lower arch, open bite, micrognathia, Microstomia, incompetent lips, caries in relation to 53, 63, 64, 74.(Fig. 4 & 5)

Investigations:

Orthopantomograph, lateral cephalogram, CT, blood investigations.

Diagnosis:

Based on the history and records, clinical features and radiographic examination, he was diagnosed as having TCS.

Discussion:

TCS affects the growth of facial bones and tissues. The severity of the syndrome varies from person to person. TCS affects the size, shape and position of ears, eyes, eyelids, cheekbones and jaws. There is no predominance of sexes or races and it consists of autosomal dominant traits with varying expression. Its phenotypic presentation is probably due to a bilateral congenital deformity involving the first and second humeral arches.[5,6] The syndrome affects the craniofacial complex, which includes the skull, eyes, ears, nasal structures, facial bones and oral tissues. The clinical picture is usually bilaterally symmetrical and is present at birth. Although our case was present bilaterally, the hypoplasia was more pronounced on the left side of the face.

TCS was classified by Franceschetti into the following categories:

Complete: Classic main features:

Incomplete: Less severe anomalies of the ear, eye, zygoma and mandible

Unilateral: Affects one side

Abortive: Lower eyelid pseudocoloboma only and zygomatic hypoplasia

Atypical form: Combined with other abnormalities not usually included in the typical syndrome.[7,8]

Diagnostic Clinical Features[9]

I. Ophthalmological

a. Anti-Mongoloid trend of cracks
b. Hypoplasia of lower eyelids and lateral folds
c. Partial absence of eyelids
d. Hypertelorism

II. Orbits

a. Hypoplastic lateral aspects of the orbit
b. Dysplastic inferior lateral orbit

III. Ears:

a. Abnormalities of the external ear
b Abnormalities of the external ear
c. Skeletal malformations of the middle ear cavity
d. Conductive hearing loss is caused by variable hypoplasia of the external auditory canals and middle ear bones.

IV. Deformation of the facial bone:

a. Hypoplasia of the malar bones Often split arches. Limited formation of residual zygomatic complex.

V. Maxilla and mandible:

a. Typically Hypoplastic
b. Variable effects on temporomandibular joints
c. Anterior open bite
d. A sharp occlusal plane

VI. Nose/mouth:

a. Respiratory distress in severely affected patients as a result of the following two factors:

Jaw hypoplasia, which tends to narrow the nasal passages. Mandibular micrognathia and posterior tongue obstruct the oropharynx and hypopharyngeal spaces.

b. Nasal deformity
c. Microstomia
d. Cleft palate with or without cleft lip
e. High arch cleft palate
f. Supplement
g. Open bite.

VI. Differential Diagnosis[10l]

· Hemifacial Microsomia
· Nager's Acrofacial Dysostosis
· Miller Acrofacial Dysostosis

Management:

The current approach to TCS clinical deformities seeks functional and aesthetic corrections and psycho social support. A multi disciplinary approach with otolaryngologists, craniofacial surgeons, ophthalmologists, speech therapists, psychologists and pediatric dentists is the most appropriate way to treat these patients.[11]

Depending on the severity of TCS, a patient may require some or all of the following procedures: conductive hearing aid, cleft palate repair, lateral wall and orbital floor repair, cheekbones, eyelid fold repair, underdeveloped jaw and jaw repair, beak nose repair operation, ear reconstruction. There are situations where teet hare missing or the patient may not be able to open the mouth. That makes dentistry quite difficult; therefore, the special skills of a pediatric dentist are required.[12]

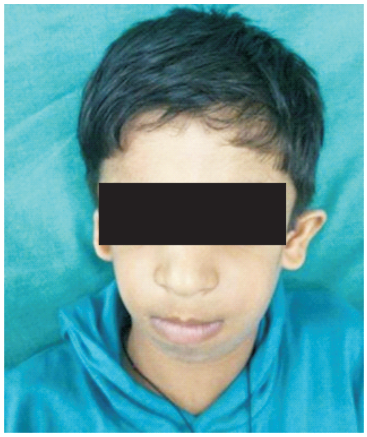

Fig 1: Profile Picture

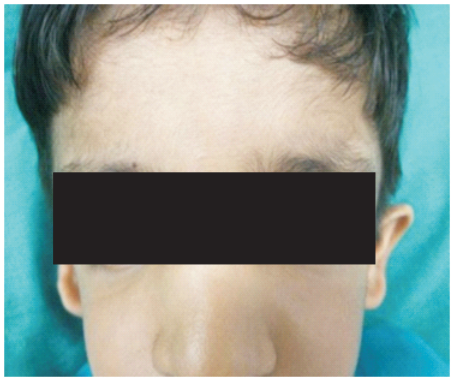

Fig 2: Broadened nasal bridge

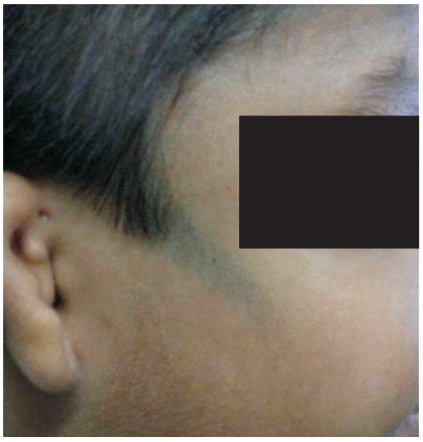

Fig 3: Facial Dysmorphic features

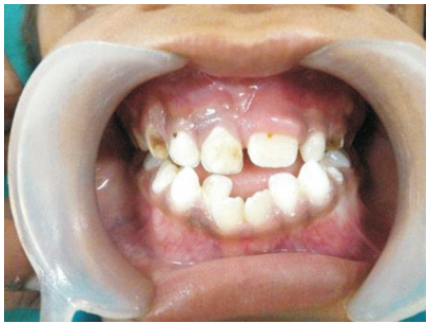

Fig 4: Malocclusion with anterior open bite

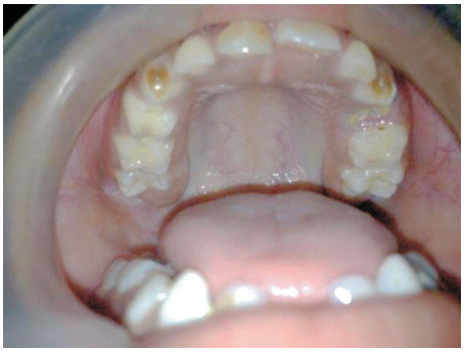

Fig 5: Intra-oral Findings

Conclusion:

Treacher Collin syndrome affects each patient differently. Surgical procedures have corrected several abnormalities of the disease. Proper planning and a conversational approach can produce excellent patient outcomes. When Treacher Collin syndrome is confirmed by clinical radiography and patient and parent history, attention to psychosocial needs is important.

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