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Case Report

Treacher Collins Syndrome - Case Report

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***ABSTRACT*:**
Treacher collins syndrome or mandibulofacial dysostosis is an autosomal dominant disorder. It is described by craniofacial development with variable phenotype expressivity. The abnormalities are derived from the first and second branchial arches. The important features are antimongoloid slanting of the palpebral fissures, depressed zygomatic bone, hypoplasia of the mandible, malformations of the auricular pinna, lower eyelid coloboma, conductive hearing loss, cleft palate. We report a case of 16 years old female with classic clinical features of this syndrome. The causes, clinical manifestations, radiographic features, differential diagnosis, and management with multidisciplinary intervention have been emphasized in the present case report.

Keywords -Treacher collins syndrome, mandibulofacial dystosis, autosomal dominant, hypoplasia

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INTRODUCTION

Treacher collins syndrome (TCS) is autosomal dominant disorder and also termed as mandibulofacial dysostosis or Treacher Collins Franceschetti syndrome. The description of the syndrome was explained by Thomson (1846) further by Toynbee (1847) and Berry (1889). Francischetti and klein in 1949 reviewed the condition and coined the term mandibulofacial dysostosis.[1] TCS is a rare genetical abnormality which leads to a developmental abnormality of facial bones, causing facial defects with large penetrance and variable expressivity.[2,3]

CASE REPORT

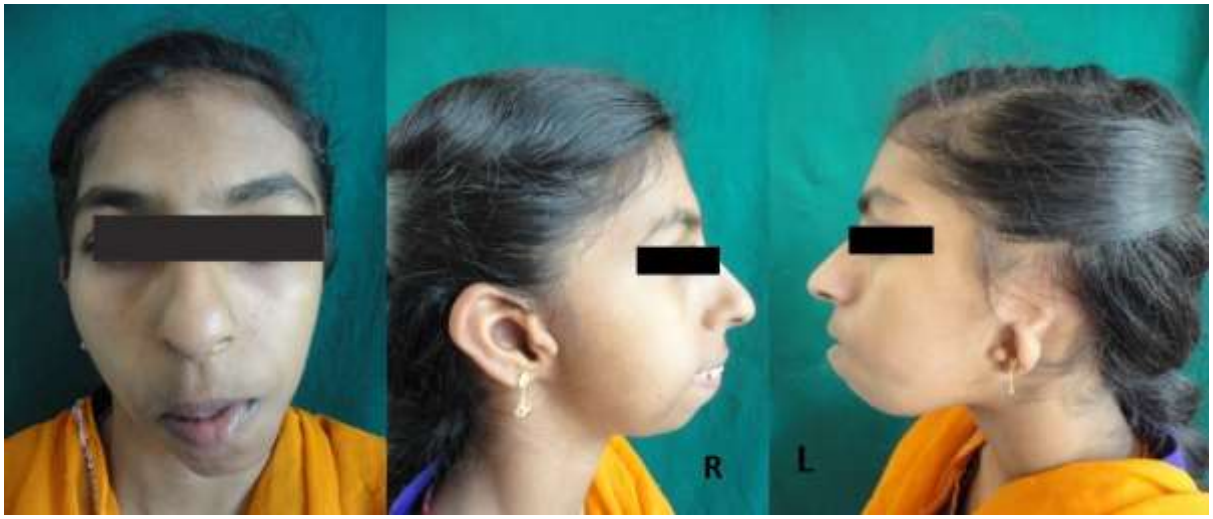
A 16-year-old female patient reported to the department of Oral Medicine and Radiology with a complaint of forwardly placed teeth in the upper front region since 6 years. Patient also gave a history of the deviated jaw and malformed ear which was present since birth. Patient had difficulty in hearing and she was not using any hearing aids. Family history was noncontributory. The patient was moderately built, nourished with normal gait. Vital signs were within normal limits.

On extraoral examination the shape of the head was dolichocephalic and shape of the face was leptoprosopic. The Left ear was small in size and placed below the level of maxilla. Gross facial asymmetry was detected on lower middle 1/3rd of the face, jaw was deviated to left side. Patient had a convex profile. Lips were incompetent. There was increased inter canthus distance. Malformed auricle present on the left side with two ear tags in number

measuring about 0.5cm which was circular. Another ear tag was present on the right ear measuring about 1 cm. There was hypoplasia of external acoustic meatus. (**Figure1)**

Nasal region was normal. Flattening cheekbones, fullness in the mentalis region with shallow mentolabial sulcus was present. Hypoplastic mandible and hypoplastic area noted from the left parasymphysis region till the angle of the mandible. The deviation was pronounced while opening the mouth and size of mouth was normal. TMJ movements were less appreciable in the left side. Flattening was present in the right zygomatic area. Depression was felt in the left inferior border of the mandible. Ramus of mandible was irregular on palpation with prominent antegonial notch on the left side.

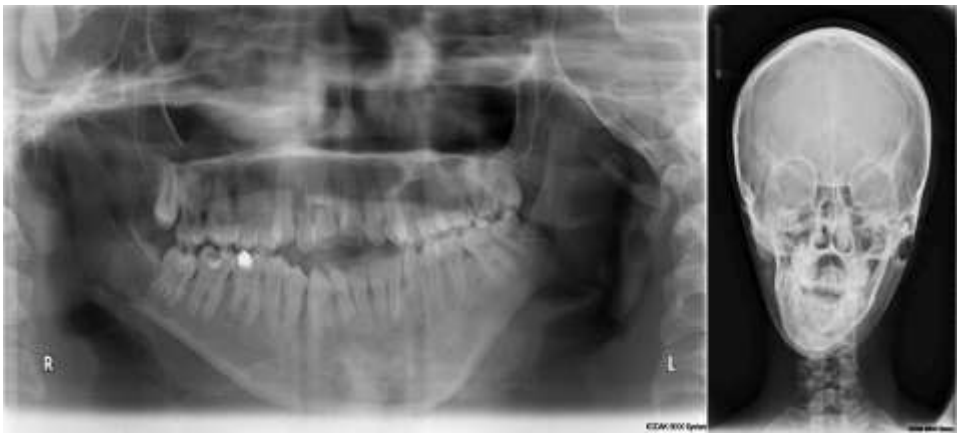
On intraoral examination there was high arched palate with anterior open bite-3mm and posterior cross bite.Orthopantogram (OPG), Posteroanterior (PA) projection and Computed tomography (CT) was advised. OPG revealed underdeveloped condyle and coronoid process, hypoplastic zygomatic arch and absence of normal mandibular ramus angle- body ratio**.** PA projection showed left condylar malformation. **(Figure 2**) 3D reconstruction of CT of shows underdeveloped condyle and short rami on the left side. **(Figure 3**) Based on the history, clinical features; and radiographic features TCS was given as diagnosis. The Patient was advised fixed Mechanotherapy with MBT appliance for unilateral arch expansion in relation to the upper left side. After alignment, Lefort 1 osteotomy was planned to correct facial asymmetry and skeletal open bite.



(a) (b) (c)

Figure 1:

a) Facial asymmetry b) tissue tags on the right ear c) tissue tag and malformed ear on left side



(a) (b)

Figure 2:

(a) OPG - underdeveloped condyle and coronoid process (b) PA view- conylar malformation of left side

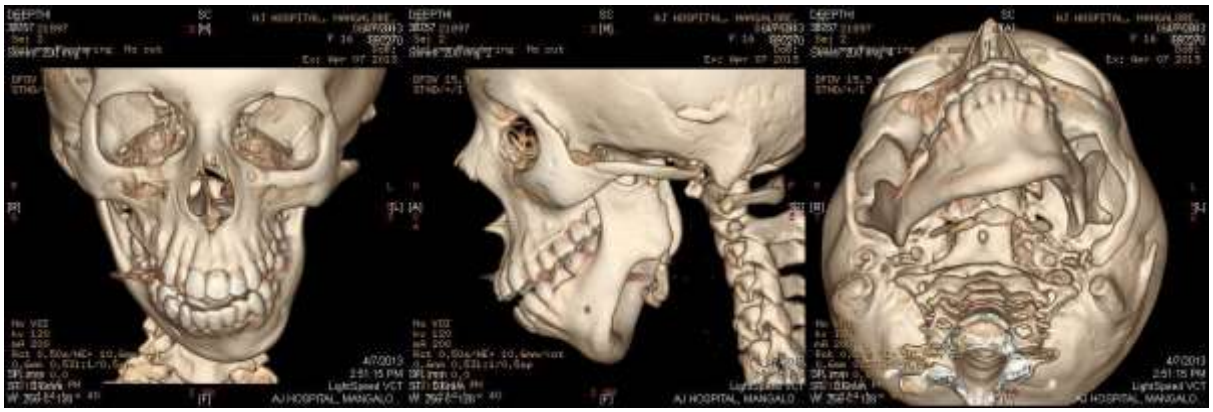


Figure 3: 3D reconstruction – Underdeveloped condyle and short rami

DISCUSSION

The TCS is a genetic disorder with developmental anomalies of the head and neck region.[4] It affects both the sex equally. The incidence of TCS is 1 in 50,000 children born alive. Around 40% of cases give family history, and the rest 60% of the cases arise because of a new mutation. The other ways of inheritance are autosomal recessive transmission, the involvement of gonadal mosaicism and chromosomal rearrangement.[5]

TCS occurs due to abnormality in the development of the first and second brachial arch. A person with TCS has almost 50% of a chance passing to his or her child.[6] Children may be affected to varying degrees when the parent with TCS passes the gene. The gene which is associated with TCS is TCOF1 which is mapped to the long arm of chromosome 5q31-q33.3 and it contains axons that encode a protein called treacle. [7]

Neural crest cells are multipotent cells which are formed in the neural ectoderm. This condition occurs due abnormal migration of neural crest cell, during development of inappropriate cellular differentiation and extracellular matrix abnormality.[5]

The clinical features of TCS vary including mild to severe symptoms. Some patients have mild features and others with facial deformities.[8] Patient will have normal intelligence level. There will be conductive hearing loss due to malformations structures present in the middle ear; tiny or absence of external ears; abnormalities of external ear like hypoplasia or lower position of the ear shells, appearance of skin extensions and blind fistula, atresia, stenosis of the external auditory canal which was also seen in our case.[2] The deformities of the nasal region are huge, beak-like nose with blocked minor nasal passageway, downward slanting of eyes, notching of lower eyelid, eye deformities include impaired vision, coloboma of the upper lid, hypertelorism, poorly developed or absent cheekbones, floor and lateral wall of the eye socket, macrostomia, high vaulted palate. Around 60% of TCS patients have tooth abnormalities like agenesis of the tooth (33%), deformities of enamel (20%), and mal-positioned maxillary first molars.[6,1]

The development of OMENS classification helped incomplete and stage-based method to classify the disorder into the complete (presenting all the known features), incomplete (less severe ear, eye, zygoma and mandibular abnormalities), abortive (only lower lid pseudo coloboma and zygomatic hypoplasia), unilateral (anomalies limited to only one side of the face), and the atypical (combined with other abnormalities not usually part of this syndrome) forms.[6]

The differential diagnosis of TCS are Nager syndrome, Millers syndrome, Goldenhar syndrome. The facial features of Nager syndrome are similar to TCS in the eye region which include downward slanting of eyes and decreased eyelashes. The abnormalities of the preaxial limb is one of the important features of Nager syndrome. Millers syndrome has characteristics like the outward turning of the eyelid margin. The limb abnormalities include postaxial with absence or partial growth of the fifth digital ray of all four limbs.[4,9] Goldenhar syndrome usually affects only one side of the face and showing deformities on the ocular and vertebra.[10] Prenatal diagnosis should be made using ultrasonography for the detection of the syndrome. Mutation of TCOF1 can be checked by amniocentesis.[4,9]

The abnormalities of TCS can be investigated by using routine radiographs or computed tomography with three-dimensional reconstruction. The important features comprise of underdeveloped or missing zygomatic arches. In contrast the body of the zygoma is the less affected area, choanal shortening, retrognathism present with smaller mandible, and tapering of the maxilla seen. The inner ear structures are not affected since they are not derived from brachial arches. The external auditory canal appears to be missing or hypoplastic. The structures of middle ear abnormalities include hypoplastic and dysmorphic middle ear space and dysfunction of ear ossicles.[8]

The individuals diagnosed with TCS needs an interdisciplinary group including craniofacial surgeons, orthodontists, ophthalmologists, otolaryngologists, and speech therapist. Some individuals may require multiple reconstructive

procedures for the correction of deformities like conduction of bone amplication for hearing aid, surgical correction of the side wall and floor of the eye socket and eyelid, cleft palate surgery and reconstruction of the poorly developed jaw and. Baha (Bone anchored hearing aid) is another alternative for hearing aid. Mutation of TCOF1 can be checked by amniocentesis.[11] Genetic advisory support is suggested for affected persons and their family members. The reconstruction of zygomatic and orbital region is done when the cranio-orbitozygomatic bone is completely developed.[6]

CONCLUSION:

TCS is a autosomal dominant disorder with many craniofacial abnormalities. Well planned treatment with multidisciplinary intervention can help the child affected with TCS. The gene responsible for TCS can be detected by prenatal scanning which helps in reducing the incidence of TCS.

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