



Case Report

Treacher collins syndrome

Anjum Ara J Farooqui^{1,*}, Anita Munde¹, Sunil Mishra¹, Jamebaseer M Farooqui²

¹Dept. of Oral Medicine & Radiology, Rural Dental College of Pravara Institute of Medical Sciences (Deemed to be University), Ahmednagar, Maharashtra, India

²Dept. of Forensic Medicine & Toxicology, Dr. Balasaheb Vikhe Patil Rural Medical College of Pravara Institute of Medical Sciences (Deemed to be University), Ahmednagar, Maharashtra, India



ARTICLE INFO

Article history:

Received 28-03-2021

Accepted 06-04-2021

Available online 29-04-2021

Keywords:

Condylar hypoplasia

Hypoplastic maxilla

Hypoplastic zygoma

Treacher Collins syndrome

ABSTRACT

Treacher Collins syndrome (TCS) or Franceschetti syndrome is an autosomal dominant disorder of craniofacial development with variable expressivity. Incidence of this syndrome is approximately 1 in 50,000 live births and it affects both genders equally. TCS affects structures which are derivatives of the first and second brachial arches which was also observed in this patient presenting with antimongoloid slanting of the palpebral fissures, colobomas of the lower eyelid, hypoplasia of zygoma and mandible, and an ear abnormality. This article describes clinical features of TCS in a 21-years-old boy.

© This is an open access article distributed under the terms of the Creative Commons Attribution License (<https://creativecommons.org/licenses/by/4.0/>) which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

1. Introduction

Treacher Collins syndrome (TCS), also called Treacher Collins-Franceschetti syndrome or mandibulofacial dysostosis, is an autosomal dominant disorder affecting the development of structures derived from the first and second brachial arches during early embryonic development. The estimated incidence of TCS ranges from 1:40,000 to 1:70,000 of live births.¹⁻³ TCS is characterized by deafness, hypoplasia of facial bones (mandible, maxilla and cheek bone), antimongoloid slant of palpebral fissures, coloboma of the lower lid and bilateral anomalies of auricle. It is a condition in which the cheek bones and jawbones are underdeveloped.³

2. Case Report

A 21-year-old boy reported to the department of oral medicine with a chief complaint of difficulty in breathing from left nostril. Patient gives history of same since birth and patient has not consulted any physician till

now. Physical examination revealed antimongoloid face with slanting of the palpebral fissures, sparse eyelashes on both lower eye lids with an inability to close left eye. There was hypoplasia of malar prominence resulting in a “sunken” appearance temporally causing nose to appear very prominent, face was depressed on the left side, underdeveloped zygoma and maxilla seen on the left side of the face, deviation of nose towards left side and slanting of the left eye lid seen. Patient also gave history of blurred vision in sunlight. Left side ear appears small as compared to right side. On intra oral examination, high arched palate was seen, over-retained (FDI) 62,82 and 83, lingually erupted 43, missing 42, 32 and 27. Panoramic findings revealed over-retained 63, 82 and 83; impacted 18, 28, 38 and 48. Altered condylar shape was seen on left side with decreased height of ramus on left side. The angle of mandible on the left side appeared sharp. CBCT findings were as follows: Skull- Brachycephalic depression was seen on the temporal bone on right side. Bilaterally elongated styloid process was seen alongwith underdeveloped mastoid air cells on left side; Maxilla- Underdeveloped zygomatic bone and maxilla on left side, wide nasopalatine canal, thin

* Corresponding author.

E-mail address: fanjumj@gmail.com (A. A. J. Farooqui).

and short hard palate with thick and elongated soft palate; Mandible- Bilateral condylar hypoplasia, underdeveloped ramus (short ramus height and obtuse gonial angle) and body of mandible on left side; Nasal findings- severe DNS towards right side with underdeveloped superior middle and inferior meatus on right side; Paranasal sinuses- Underdeveloped frontal sinus on right side, underdeveloped maxillary sinus on left side, bilateral septa seen in maxillary sinuses; Dental findings- over retained 63, 82 and 83, palatally impacted 23, mesioangularly impacted 38 and 48, vertically impacted 28 and 18, missing 27, 32 and 42. Based on the clinic-radiographic features a working diagnosis of Treacher Collin syndrome was given. The patient was further referred to ENT department for evaluation and management of his chief complaint.



Fig. 3:



Fig. 1:

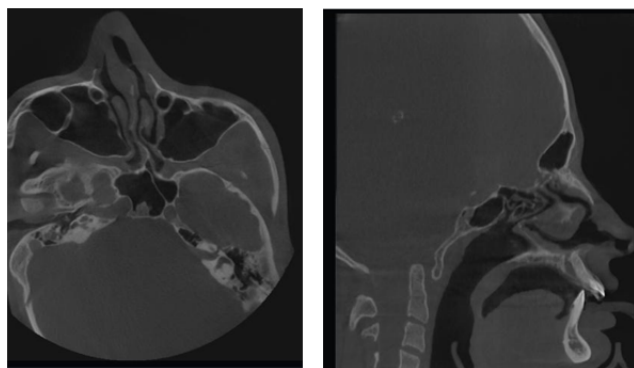


Fig. 2:

3. Discussion

TCS or Franceschetti syndrome, is an autosomal dominant disorder of craniofacial development.^{4,5} Pathogenesis of TCS has been postulated to be due to several factors such as abnormal patterns of neural crest cell migration, abnormal domains of cell death, improper cellular differentiation during development, or an abnormality of the extracellular

matrix.^{4,6} It affects both genders equally and has a family history in 40% of cases while the remaining 60% appears to arise as a result of a de novo mutation.^{4,7} There was no family history in our case. Derivatives from the first and second pharyngeal arches, grooves, and pouches are affected in TCS.^{8,9} The typical characteristics of the TCS as stated by Franceschetti and Klein¹⁰ in 1949 are as follows: (1) Antimongoloid palpebral fissures with either a notch or coloboma of outer third of the lower lid and occasional absence or paucity of lashes of lower eyelid. Antimongoloid palpebral fissures were observed in the reported case on the left side.² Hypoplasia of facial bones, especially the malar bones and mandible. These features were present in our case.³ Malformation of the external ear and occasionally middle and inner ear, with low implantation of the auricle. Ear malformation on the left side was observed in our case.⁴ Macrostomia, high-arched palate, malocclusion, and abnormal position of the teeth. Although macrostomia was not observed but high arched palate and malocclusion was present in our case.⁵ Association at times with other anomalies, such as obliteration of the naso-frontal angle, and ear and skeletal deformities. Obliteration of the nasofrontal angle and cleft was observed in our case. Most of the clinical features in a patient presenting with TCS were found in our case which gives it the merit of a classic case of TCS. A multidisciplinary approach is required for the management of individuals affected by TCS involving craniofacial surgeons, orthodontists, ophthalmologists, otolaryngologists, and speech therapists.¹ In this case, patient had difficulty in breathing and was referred to ENT department for further management.

4. Conclusion

TCS is an autosomal dominant disorder of craniofacial development which presents with unusual clinical features associated with abnormalities of structures derived from the first and second brachial arches, including antimongoloid slant of palpebral fissures, colobomas of the lower eyelid,

eyelash malformations, deafness, hypoplasia of facial bones (mandible, maxilla and cheek bone) and malar and mandibular defects. Management of TCS needs a multidisciplinary approach. The treatment plan is made to meet the individual patients need, considering the growth patterns, function and psychological development.

5. Source of Funding

None.

6. Conflict of Interest

The authors declare that there is no conflict of interest.

References

1. Marszałek B, Wójcicki P, Kobus K, Trzeciak WH. Clinical features, treatment and genetic background of Treacher Collins syndrome. *J Appl Genet.* 2001;43(2):223–33.
2. Singh G, Garg A, Awana RS, Kumar A, Agarwal A, Walia N, et al. Treacher Collins Syndrome: A Case Report and Review of Literature. *Int J Oral Health Med Res.* 2017;4(3):67–9.
3. Shetty SB, Pidamale R. Treacher Collins Syndrome: A Case Report and a Brief Review on Diagnostic Aids. *Int J Clin Pediatr Dent.* 2011;4(3):235–9. doi:10.5005/jp-journals-10005-1116.
4. Sumbh SG, Pagare J, Sumbh B. Treacher collins syndrome - Report of a classical case. *J Cleft Lip Palate Craniofac Anomal.* 2017;4(1):69–72. doi:10.4103/2348-2125.205412.
5. Trainor PA, Dixon J, Dixon MJ. Treacher Collins syndrome: etiology, pathogenesis and prevention. *Eur J Hum Genet.* 2009;17(3):275–83. doi:10.1038/ejhg.2008.221.
6. Agrawal SM, Parihar SS, Agrawal MG. Mandibulofacial dysostosis (Treacher Collins syndrome)-A rare case report. *Natl J Med Dent Res.* 2013;1:34–7.
7. Kasat V, Baldawa R. Treacher Collins syndrome-a case report and review of literature. *J Clin Exp Dent.* 2011;3(1):e395–9. doi:10.4317/jced.3.e395.
8. Hertle RW, Ziylan S, Katowitz JA. Ophthalmic features and visual prognosis in the Treacher-Collins syndrome. *Br J Ophthalmol.* 1993;77(10):642–5. doi:10.1136/bjo.77.10.642.
9. Farrar JE. Mandibulo-facial dysostosis. A familial study. *Br J Ophthalmol.* 1967;51(2):132–5. doi:10.1136/bjo.51.2.132.
10. Alva P, Shetty SR, Babu SG, Madiyal A. Treacher Collins syndrome - a report of two cases with review of literature. *El Mednifico J.* 2015;2(4):428–9. doi:10.18035/emj.v2i4.208.

Author biography

Anjum Ara J Farooqui, Reader

Anita Munde, Professor and HOD

Sunil Mishra, Associate Professor

Jamebaseer M Farooqui, Professor

Cite this article: Farooqui AAJ, Munde A, Mishra S, Farooqui JM. Treacher collins syndrome. *IP Int J Maxillofac Imaging* 2021;7(1):25-27.