

Case Report

Case report of Treacher–Collins syndrome with a review of its pathogenesis

Arijita Paul¹, Debasish Pramanick², Prateeti Ghosh¹Departments of ¹Oral and Maxillofacial Surgery, ²Oral Pathology and Microbiology, Burdwan Dental College and Hospital, East Burdwan, West Bengal, India.

ABSTRACT

Treacher–Collins syndrome (TCS) is a rare autosomal-dominant orofacial anomaly which is also known as Franceschetti–Zwahlen–Klein syndrome and mandibulofacial dysostosis. Edward Treacher Collins, a renowned British ophthalmologist, first illustrated this orofacial anomaly in 1900. This craniofacial dysmorphic disorder shows features such as hypoplasia of the malar bone and mandible, downward slanting of the lower palpebral fissure, external ear malformation, coloboma of the eyelids, dental abnormalities like malocclusion, and open bite. Pathogenesis of TCS is due to a mutation of the TCOF1 gene. As a result of the mutation of the TCOF1 gene, there is a defect in the formation of the phosphoprotein treacle. This phosphoprotein is very much related to the proliferation and survival of cephalic neural crest cells. As there is a defect in treacle, there is an altered function of neural crest cells which ultimately leads to the manifestation of TCS. Here, we report a case of TCS in a 21-year-old female patient.

Keywords: Coloboma, Mandibulofacial dysostosis, *TCOF1* gene, Malocclusion, Treacher- Collins syndrome

INTRODUCTION

Treacher–Collins syndrome (TCS) is a rare orofacial anomaly which is also known as Franceschetti–Zwahlen–Klein syndrome and mandibulofacial dysostosis. In the year 1846, Thomson presented this disorder for the 1st time, and later in 1889, George Andreas Berry designated it as a congenital deformity with the presence of coloboma of the lower eyelids. Edward Treacher Collins, a renowned British ophthalmologist, first illustrated this orofacial anomaly in 1900. Pathogenesis of TCS is related to the aberrations occurred in the embryonal craniofacial structures derived from the first and second branchial arches during their histodifferentiation and morphogenesis stage between the 20th day and 12th week of intrauterine life.^[1] Literature review shows that the estimated incidence of TCS is about 1 among 50000 live births.^[2] Nearly 60% cases of TCS appear as *de novo* mutations.

Clinically, the affected individuals exhibit underdevelopment of cheekbones, jaws, and palate, along with downward slanting of palpebral fissures. Anomalies of the external and middle ear often leading to partial or complete hearing loss are also seen.^[3] Microcephaly, behavioral disorders, and psychomotor delay are also reported in TCS.^[4]

Dental anomalies, namely malocclusion, misaligned teeth, enamel hypoplasia, spacing, hypodontia, and anterior open bite, are seen in TCS. High arched palate with or without palatal cleft is also reported in many cases of TCS. Patients with TCS may exhibit mental retardation or normal intelligence level. However, the appearance of the patients may lead to social challenges and social stigma.

CASE REPORT

A 21-year-old female patient reported to the Department of Oral and Maxillofacial Pathology of a tertiary health care center with a complaint of pain in the right lower back tooth region. After recording proper history, extraoral clinical examination was performed, which revealed a narrow, convex facial profile with mandibular and zygomatic hypoplasia [Figure 1a] along with antimongoloid slanting of both eyes [Figure 1b]. Eyelashes in the lower eyelid were partially absent. The maxilla was constricted with protrusion of the upper anterior teeth. There was “sunk-in” appearance of the face, causing the nose to be very prominent. Bilateral microtia with rudimentary pinna was also noted [Figure 1c]. In addition, atresic external ear canals and absence of external ear opening were also noted. The patient had deafness with a 60% reduction in hearing. Intraoral examination showed

*Corresponding author: Debasish Pramanick, Department of Oral Pathology and Microbiology, Burdwan Dental College and Hospital, East Burdwan, West Bengal, India. dr.pramanick.debasish@gmail.com

Received: 25 December 2025 Accepted: 25 February 2026 Published: 27 April 2026 DOI: 10.25259/SRJHS_26_2025

This is an open-access article distributed under the terms of the Creative Commons Attribution-Non Commercial-Share Alike 4.0 License, which allows others to remix, transform, and build upon the work non-commercially, as long as the author is credited and the new creations are licensed under the identical terms. ©2026 Published by Scientific Scholar on behalf of Sri Ramachandra Journal of Health Sciences



Figure 1: A 21-year-old female patient presented with features of Treacher–Collins syndrome. (a) Frontal profile of the patient showing dental malocclusion, (b) antimongoloid slanting of both eyes with partial absence of eyelashes in the lower eyelid, (c) ear deformity, (d) high-arched palate, (e) lateral cephalogram of the patient showing mandibular hypoplasia, prominent antegonial notches, open bite, and convex facial profile.

“V” shaped, narrow, high-arched palate [Figure 1d], crowded alignment of teeth in both arches, multiple carious teeth (11, 36, 37, 46), retained root of deciduous tooth (52), and macroglossia [Figure 1a]. The patient had an open bite and a history of cleft palate. The cleft palate was repaired at the age of 2 years in the patient. Prominent antegonial notch and “double-chin” appearance were also found. The patient had a sibling but he had no such abnormalities. At the time of her birth, the paternal and maternal ages were 30 years and 27 years, respectively. There was no familial history of this disorder. No psychomotor delay or behavioral abnormalities were reported.

Lateral cephalogram of the patient was done and it revealed mandibular hypoplasia, prominent antegonial notches, open bite, and convex facial profile [Figure 1e].

Based on the clinical and radiological findings, a diagnosis of TCS was made.

DISCUSSION

TCS or mandibulofacial dysostosis is one of the autosomal dominant disorders with incomplete penetrance and variable expressivities.^[5] Pathogenesis of TCS may be attributed to the responsible gene *TCOF1* which is located on chromosome no 5 at q31 and 3q32. This gene was identified in 1996.

51 mutations in the *TCOF1* gene have been detected.^[6] As a result of mutation of the *TCOF1* gene, there is a defect in the production of nuclear phosphoprotein “Treacle.” This phosphoprotein is very much related to the proliferation and survival of cephalic neural crest cells. Various kinds of mutations such as insertions, deletions, and interpositions have been reported in various regions of the *TCOF1* gene. Only 40% of cases show positive family predilection of this syndrome. The adult patient with features of TCS shows a convex facial appearance, an accentuated and prominent nose, hypoplastic mandible and underdeveloped chin, antimongoloid slanting of palpebral fissures, coloboma with hypoplasia of lower eyelids, and lateral canthi together with partial absence of eyelashes. Tongue-shaped hair process extending into the preauricular region, malformation of the external ear, impaired or decreased hearing ability due to hypoplasia of external auditory canals, and middle ear ossicles are prominent features of TCS. Often, due to constriction of the upper respiratory tract, respiratory distress may develop in some patients. In about 35% of cases, microstomia with a narrow palatal vault and cleft palate is seen.^[6] In this present case, cleft palate with anterior open bite was present. The cleft palate was repaired at 2 years of age of patient. Other dental anomalies such as supernumerary teeth, enamel

opacity with hypoplasia, anodontia, microdontia, rotated tooth, and ectopic tooth are seen. Detailed patient history, thorough clinical extra and intraoral examination, and specialized imaging techniques such as lateral cephalogram, orthopantomogram, and CT scan may be performed for proper diagnosis and to assess the extent of craniofacial abnormalities.^[7] Jahrsdoerfer *et al.*^[8] stated that CT scan of the temporal bone in a TCS patient shows some radiographic findings such as absence of mastoid pneumatization, ossicular disjunction, and presence of bony cleft in the lateral aspect of the temporal bone and anterior to the mastoid. Literature review shows hypoplastic ear ossicles, hypoplastic epitympanum, and even in some cases, complete absence of the middle ear and external auditory canal. Molecular genetic testing should be done to detect mutations in *TCOF1*, *POLRIC*, and *POLRID* genes.

There are various differential diagnoses of TCS such as Goldenhar syndrome, Miller syndrome, and Nager syndrome. Goldenhar syndrome includes hemifacial microsomia along with vertebral abnormalities unlike TCS. Miller syndrome exhibits additional features of ectropion of the lower eyelids. Nager syndrome involves malformation of the forelimbs along with facial abnormalities.

Ultrasound screening and anomaly scan during the mid-late gestation period help to detect severe abnormalities in a case of TCS. As far as the treatment of TCS patients is concerned, a multidisciplinary approach is required, which includes reconstructive maxillofacial surgery of cheekbones along with external ear prosthesis.^[9-11] Orthognathic surgery, distraction osteogenesis together with post-operative orthodontic management, not only helps to provide a good quality of life to a TCS patient but also gives a proper esthetic appearance and functional ability.^[9]

CONCLUSION

Dental practitioners must be aware of these manifestations and should be able to recognize the disorder. Early diagnosis and correct treatment planning with regular follow-up are required to provide the quality of life for the patient. Besides that, psychological counseling of both the patient and parents is required.

Authors' contributions: AP, DP, PG: Conceptualization, methodology, software, validation, formal analysis, investigation, resources, data curation, writing - original draft, writing - review & editing, visualization, supervision, project administration, funding acquisition.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given consent for their images and other clinical information to be reported in the journal. The patient understands that the patient's names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship: Nil.

Conflicts of interest: There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation: The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

- Magalhaes MH, Da Silveria CB, Moreira CR, Cavalcanti MG. Clinical and imaging correlations of Treacher Collins syndrome: Report of two cases. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 2007;103:836-42.
- National Organization for Rare Disorders. Rare Disease Database; 1999. Available from: <https://rarediseases.org/rarediseases/treacher-collins-syndrome> [Last accessed on 2025 Dec 18].
- Poswillo D. The pathogenesis of the treacher collins syndrome (mandibulofacial dysostosis). *Br J Oral Surg* 1975;13:1-26.
- Teber OA, Gillessen-Kaesbach G, Fischer S, Böhringer S, Albrecht B, Albert A, *et al.* Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. *Eur J Hum Genet* 2004;12:879-90.
- Dalben GD, Costa B, Gomide MR. Prevalence of dental anomalies, ectopic eruption and associated oral malformations in subjects with Treacher Collins syndrome. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 2006;10:588-92.
- Marszałek B, Wojcicki P, Kobus K, Trzeciak WH. Clinical features, treatment and genetic background of Treacher Collins syndrome. *J Appl Genet* 2002;43:223-33.
- Posnick JC. Treacher Collins syndrome: Perspectives in evaluation and treatment. *J Oral Maxillofac Surg* 1997;55:1120-33.
- Jahrsdoerfer RA, Yeakley JW, Aguilar EA, Cole RR. Treacher Collins syndrome: an otologic challenge. *Ann Otol Rhinol Laryngol* 1989;98: 807-12.
- National Organization for Rare Disorders. The Physicians Guide to Treacher Collins Syndrome. Danbury, Connecticut: National Organization for Rare Disorders; 2012.
- Martelli-Junior H, Coletta RD, Miranda RT, Barros LM, Swerts MS, Bonan PR. Orofacial features of Treacher Collins syndrome. *Med Oral Patol Oral Cir Buccal* 2009;14:E344-8.
- Mittman DL, Rodman OG. Mandibulofacial dysostosis (Treacher Collins syndrome): A case report. *J Natl Med Assoc* 1992;84:1051-4.

How to cite this article: Paul A, Pramanick D, Ghosh P. Case report of Treacher–Collins syndrome with a review of its pathogenesis. *Sri Ramachandra J Health Sci.* 2026;6:58-60. doi: 10.25259/SRJHS_26_2025