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Case Report

Imaging of Treacher Collins syndrome: A case report ☆

Abhikanta Khatiwada[a,*], *Bikram Thapa*[a], *Raju Pandit*[a], *Dependra Bhandari*[b], *Sharada KC*[c]

[a] *Department of Radiology; Tribhuvan University Teaching Hospital, Kathmandu, Nepal*
[b] *Department of Orthopedic Surgery, KMCTH, Kathmandu, Nepal*
[c] *Department of Internal Medicine, NAIHS, Kathmandu, Nepal*

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ABSTRACT

Treacher Collins syndrome, also known as mandibulofacial dysostosis, is a rare congenital disorder affecting craniofacial development. It is caused by an autosomal dominant mutation, primarily in the TCOF1 gene, which impacts the development of the first and second branchial arches. We present the case of a 12-year-old male with bilateral conductive hearing loss and deformed ears, whose clinical and imaging findings were consistent with Treacher Collins syndrome. Imaging revealed microtia, atresia of the external auditory canals, and hypoplastic middle ear structures. Additionally, facial abnormalities such as retrognathia, hypoplastic zygomatic bones, and a cleft palate were identified. This case underscores the crucial role of imaging in diagnosing Treacher Collins syndrome and guiding multidisciplinary management strategies.

Introduction

Treacher Collins syndrome (TCS), also known as mandibulofacial dysostosis, is a rare congenital anomaly with an estimated incidence of about 1 in 50,000 live births [1]. The syndrome was named after Edward Treacher Collins (1862-1932), an English ophthalmologist and surgeon, who described many of its features [1]. TCS is caused by an autosomal dominant genetic mutation, leading to bilateral malformations of the first and second branchial arches [1]. The primary genes involved in its development include TCOF1, which is most commonly affected, along with POLR1C and POLR1D [1,2]. Craniofacial abnormalities in TCS typically involve the zygomatic bones, jaws, palate, and oral cavity, which can result in respiratory and feeding difficulties [3]. Conductive hearing loss is a common presenting symptom due to deformities in the external and middle ear.

We present a case of a 12-year-old male with clinical features consistent with TCS. Imaging was advised for further evaluation, and the findings confirmed the diagnosis.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

* Corresponding author.
E-mail address: avikant12@gmail.com (A. Khatiwada).

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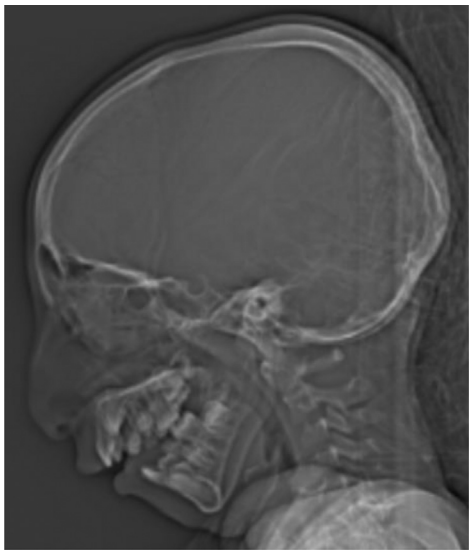

Fig. 1 – Computed tomography (CT) scannogram showing retrognathia, hypoplastic mandible (micrognathia), and an obtuse mandibular angle.

Case presentation

A 12-year-old male presented with bilateral deformed external ears and conductive hearing loss since childhood. All other developmental milestones were normal. The patient had facial disfigurement, including small cheeks. There was no significant family history of similar conditions. His mother did not report any history of teratogenic drug intake during pregnancy. On examination, the patient was found to have bilateral microtia with canal atresia. Clinically, the head was dolichocephalic, with antimongoloid slanting palpebral fissures and sparse eyelashes. Auscultation of the heart revealed normal heart sounds. Audiometric testing showed bilateral severe conductive hearing loss. Despite these challenges, the patient's IQ and speech performances were within the normal range. Based on these clinical findings, a provisional diagnosis of TCS was made, and imaging was recommended for confirmation and further management. The differential diagnosis included Auriculocondylar syndrome, Goldenhar syndrome, and Nager syndrome.

A computed tomography (CT) scan of the face and temporal bones (Figs. 1–4) revealed the absence of both the bony and cartilaginous portions of the external auditory canal, along with deformed pinnae. Bilateral middle ear cavities appeared hypoplastic with absent ossicles, and bilateral mastoids were hypoplastic and poorly pneumatized. The inner ears were grossly normal. Additional findings included a downward slant of the superior orbital walls, hypoplastic facial bones, and hypoplastic bilateral zygoma with poorly formed zygomatic arches. There was crowding of the teeth in both jaws. The condylar and coronoid processes of the mandible were normal. A small cleft palate was noted on the posterior hard palate, with normal bilateral choanae. There was a retrognathia, a hypoplastic mandible (micrognathia), and an obtuse mandibular angle. Left-sided deviation of the nasal septum with a bony spur was also observed. The visible portions of the paranasal sinuses were normal. Based on these clinical and imaging findings, a diagnosis of TCS was made.

The patient's parents declined genetic testing due to financial limitations. The patient was scheduled for the placement

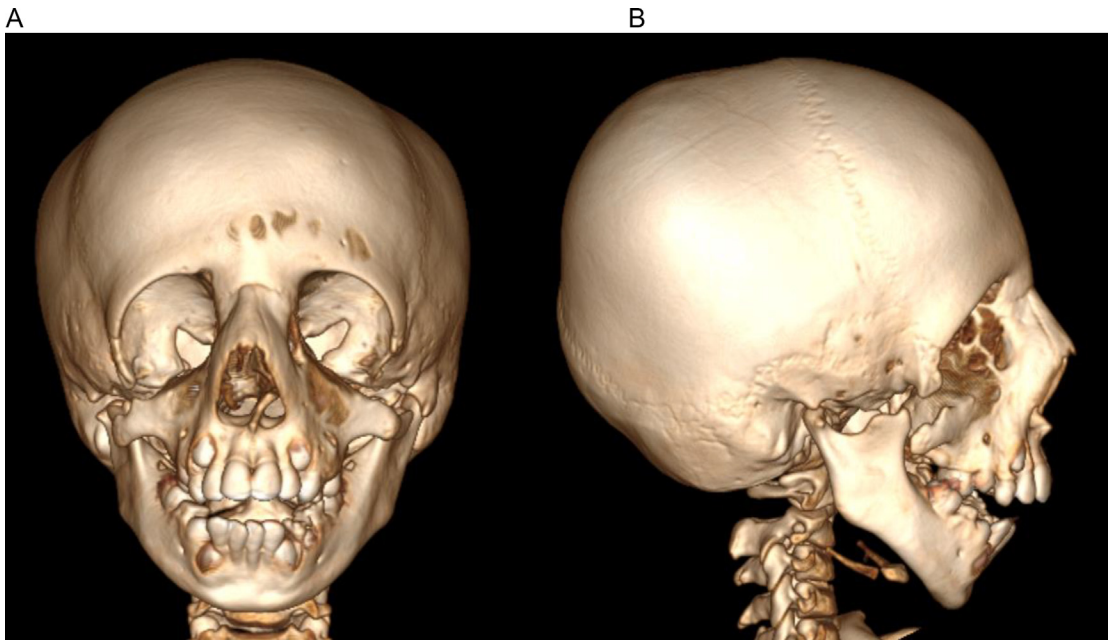

Fig. 2 – (A and B) 3D CT Volume Rendering Technique (VRT) images – Frontal and lateral views showing downward slant of the superior orbital walls, hypoplastic facial bones, hypoplastic bilateral zygoma with poorly formed zygomatic arch, micrognathia, and retrognathia. Crowded teeth are also noted in both jaws.

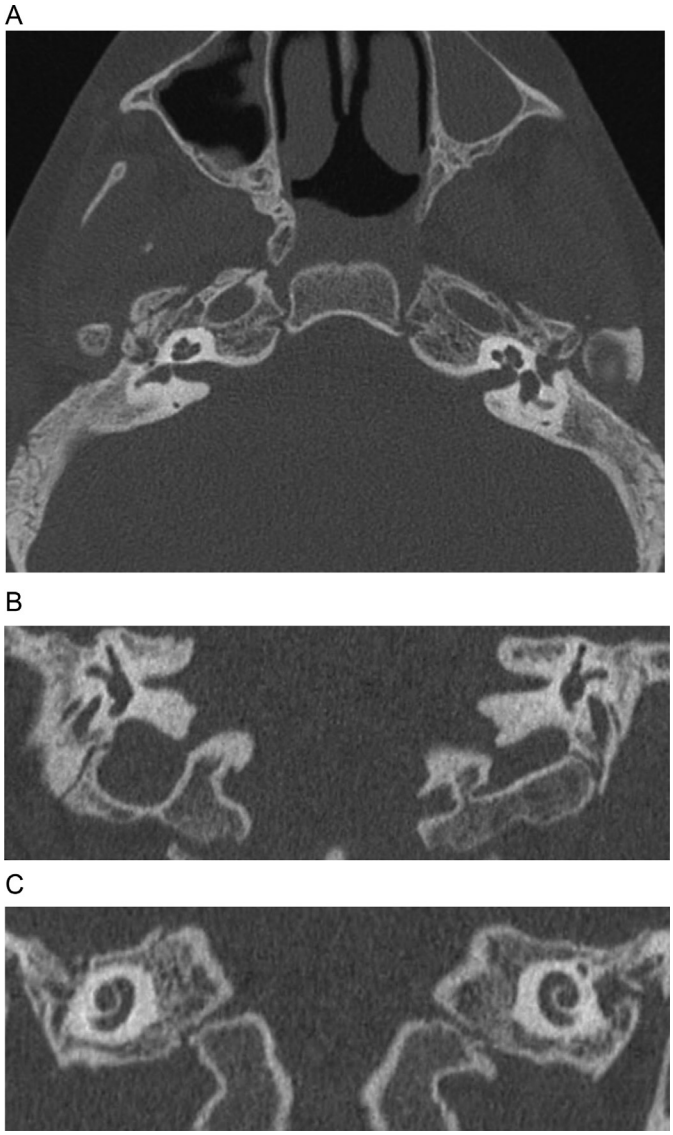

Fig. 3 – (A-C) High resolution CT (HRCT) scan of the temporal bone in axial and coronal sections revealing the absence of both the bony and cartilaginous portions of the external auditory canal. The bilateral middle ear cavities are hypoplastic with absent ossicles. Bilateral mastoids are hypoplastic and poorly pneumatized. The bilateral inner ear structures appear grossly normal.

of a bone-anchored hearing aid to correct the conductive hearing loss.

Discussion

In TCS, imaging studies typically reveal distinct craniofacial abnormalities that are often bilateral and symmetric. Common findings in the dental and mandibular regions include retrognathia and micrognathia, where the jaw is underdeveloped, leading to a receding chin. Hypoplasia or aplasia of the condylar and coronoid processes of the mandible may also be present. Additionally, there may be marked bowing of the lower border of the mandible and a concave curvature of the horizontal ramus, which is considered pathognomonic for the syndrome. The maxilla may be narrow, often accompanied by an elevated or narrow palate. Associated findings may include cleft palate, dental malocclusion, and macrostomia. The zygomatic arch is frequently malformed, underdeveloped, or absent, contributing to facial asymmetry [1,3,4].

The findings in the ear include microtia (small or malformed external ears) and aplasia of the external auditory meatus. The middle ear structures may also be involved, with hypoplasia or aplasia of the ossicles and the middle ear cavity, contributing to conductive hearing loss [1,3,4].

Ocular findings in TCS include downward slanting palpebral fissures, often due to a lack of support from the underdeveloped zygoma. The lower inner third of the eyelids may also be absent. Other ocular anomalies can include absent eyelashes, notched irises, notched choroid, and colobomas.

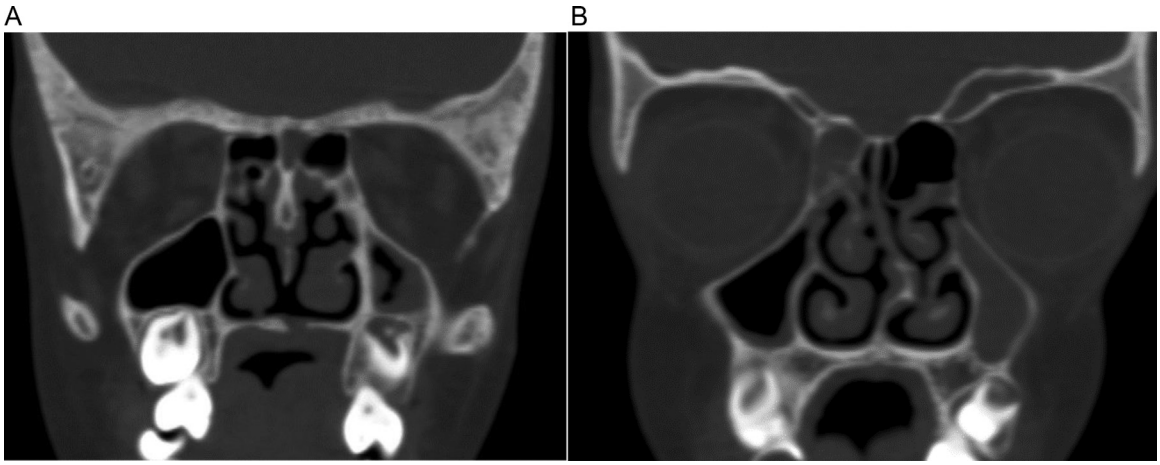

Fig. 4 – (A and B) Coronal CT images showing a small cleft palate in the posterior hard palate. There is left-sided deviation of the nasal septum with a bony spur. Bilateral paranasal sinuses appear normal.

Nasal abnormalities, such as obliteration of the nasofrontal angle leading to narrow nares, are also commonly seen. Hypoplasia of the alar cartilages and paranasal sinuses, along with choanal atresia, may be present in some cases [1,3,4]. Reduced upper airway volume and severe narrowing of the retroglossal space can result in apnea, along with speech and hearing difficulties [5].

Differential diagnoses for TCS include Auriculocondylar syndrome, Goldenhar syndrome, and Nager syndrome. Auriculocondylar syndrome presents with prominent malformed auricles, often described as "question mark ear," and specific mandibular condyle hypoplasia or aplasia [1]. In contrast, Goldenhar syndrome typically has ear anomalies, such as preauricular tags and otic hypoplasia, along with ocular anomalies like unilateral microphthalmia or epibulbar dermoids, and skeletal issues like hemifacial microsomia and vertebral anomalies [6]. Nager syndrome involves similar facial features to TCS, including mandibular hypoplasia and micrognathia, but it also includes skeletal anomalies, such as radial hypoplasia, thumb malformations, and syndactyly, which are not present in TCS [7].

Management of TCS requires a multidisciplinary approach, including reconstructive surgery to address facial abnormalities, bone conduction hearing aids or surgical correction for conductive hearing loss, and speech therapy for language development [4,8].

Conclusion

Clinical evaluation, along with correlated imaging findings, is crucial for the diagnosis of TCS. Moreover, imaging provides essential insights for surgical planning and the management of associated impairments, aiding in a comprehensive treatment approach.

Patient consent

Written informed consent was obtained from the patient's parents for publication of this case report and any accompanying images.

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