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Case Report

Treacher Collins Syndrome: A case report and literature review

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Abstract

Treacher Collins Syndrome (TCS) is a rare congenital craniofacial disorder primarily affecting the development of facial bones and tissues. It presents with characteristic features such as downward-slanting palpebral fissures, hypoplasia of the zygomatic complex and mandible, ear deformities, and conductive hearing loss. Despite its distinct facial appearance, TCS does not affect intelligence, highlighting the importance of looking beyond facial differences. This case report presents a rare instance of TCS in a young individual, detailing the clinical presentation, radiographic findings, multidisciplinary management, and psychosocial considerations. Emphasis is placed on the need for early diagnosis, genetic counseling, and long-term rehabilitative care to improve function and quality of life. The report also underscores the social and emotional challenges faced by individuals with TCS, reinforcing that they are more than a face; individuals with potential, resilience, and the right to dignity and inclusion.

Keywords: Treacher collins syndrome, Craniofacial dysostosis, Mandibulofacial dysostosis, Congenital anomaly, Case report, Psychosocial impact.

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1. Introduction

Treacher Collins Syndrome (TCS), also known as mandibulofacial dysostosis, is a rare hereditary disorder characterized by distinctive abnormalities of the craniofacial region. It follows an autosomal dominant inheritance pattern and occurs in approximately 1 in 50,000 live births. Notably, about 60% of cases arise due to spontaneous genetic mutations with no prior family history.[1] TCS mainly occurs due to alterations in the TCOF1, POLR1C, or POLR1D genes.[2] These genes play crucial roles in the development of facial skeletal elements and soft tissue structures. Significant structural abnormalities result from improper formation of the first and second pharyngeal arches during histodifferentiation and morphogenesis, occurring between approximately the 20th day and the 12th week of intrauterine life.[3]

The condition was first described by Thomson in 1846 and later defined as a congenital malformation with lower eyelid colobomata by George Andreas Berry in 1889. Edward Treacher Collins, an ophthalmologist, provided detailed illustrations of its key features in 1900, giving the syndrome its eponym. The disorder is also known as Franceschetti–Zwahlen–Klein syndrome (Franceschetti and Klein) or mandibulofacial dysostosis (MFD).[4]

Clinical manifestations are typically symmetrical and include external ear deformities such as microtia or anotia, often leading to conductive hearing loss; downslanting palpebral fissures, frequently associated with lower eyelid colobomas; hypoplasia of the zygomatic complex and mandible (micrognathia); and, occasionally, choanal atresia or cleft palate.[5] Although TCS is uncommon, its impact on affected individuals and their families can be profound, underscoring the importance of a comprehensive understanding of its etiology, clinical features, diagnosis, and management.

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2. Case Report

A 21-year-old male presented to the Department of Oral Medicine and Radiology, K.D. Dental College and Hospital, Mathura, with a complaint of pain in the lower right posterior teeth region for the past 3–4 months. The patient had been asymptomatic until four months ago, after which he began experiencing dull and continuous pain in the mandibular right posterior region.

Past dental history revealed that the patient had undergone orthognathic surgery at the age of 11 years. Past medical history was significant for cleft palate repair at the age of 2 years and septorhinoplasty performed a few months prior. Family and personal histories were non-contributory.General examination showed an endomorphic build with normal gait, stature, height, and weight. Vital signs were within normal limits.

2.1. Extraoral examination

The patient exhibited facial deformities, including marked hypoplasia of the malar region, micrognathia, and macrostomia. The eyes demonstrated lateral downslanting of the palpebral fissures (antimongoloid), with colobomas of the lower eyelids and sparse eyelashes. The right ear was malformed, low-set, and associated with stenosed external auditory canals. The patient presented with hyponasal speech. Cognitive assessment indicated normal intellect. (**Figure 1A-D**)

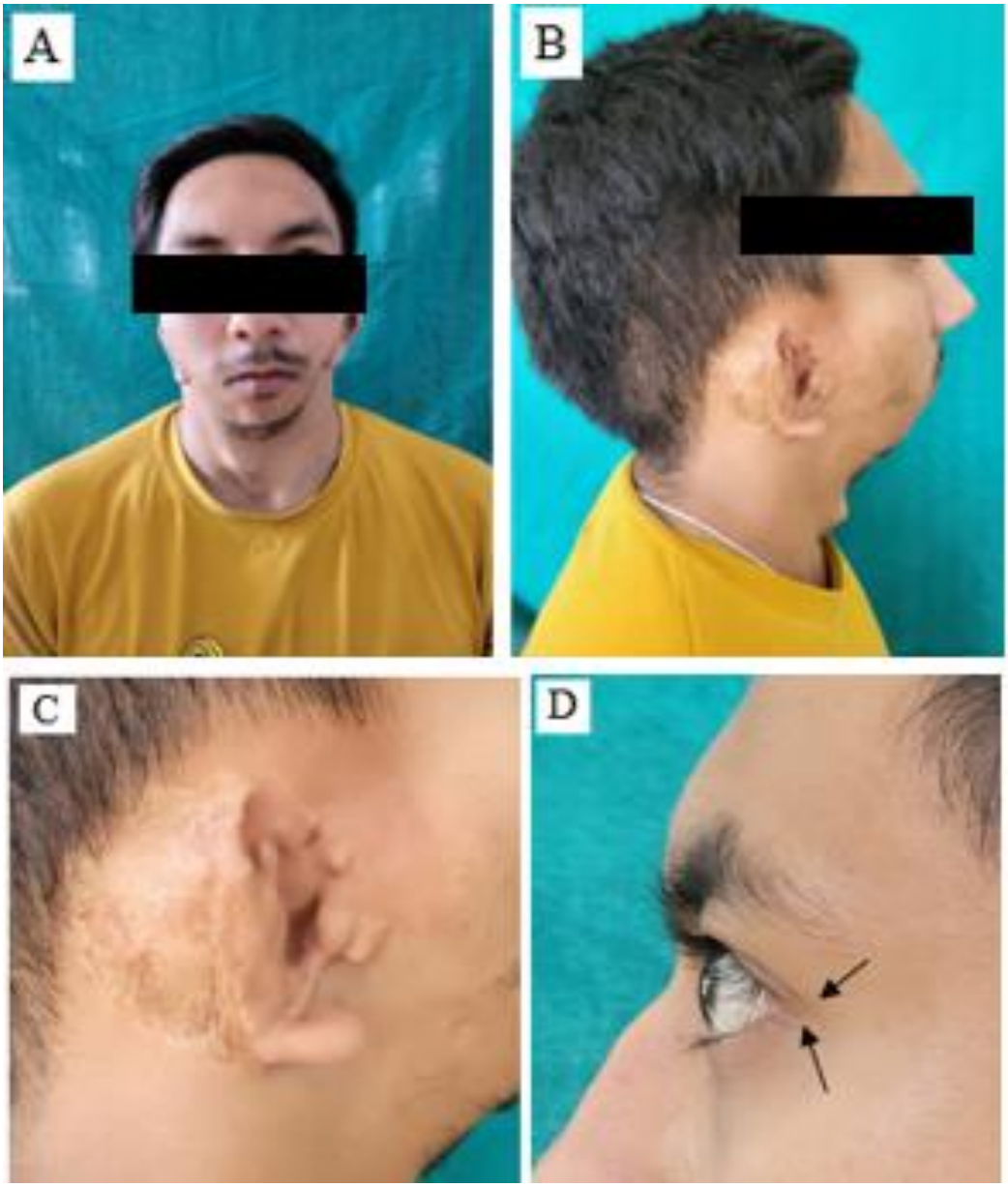

Figure 1: Extra-oral photographs depicts patient's; **A:** frontal; **B:** lateral profile; **C:** deformed ear; **D:** coloboma of eyelid.

2.2. Intraoral examination

Intraoral examination revealed a hypoplastic maxilla and mandible. All permanent canines were absent from the occlusion. A high-arched palate was observed, and the patient demonstrated fair oral hygiene.

2.3. Radiographic investigations

Radiographic evaluation included orthopantomogram (OPG), lateral cephalogram, and posteroanterior (PA) skull view.

The orthopantomogram showed a prominent antegonial notch, reduced ramus height and width, bilateral coronoid aplasia, and plating from previous mandibular osteotomy. (**Figure 2A**) The lateral cephalogram demonstrated hypoplasia of the mandible and zygomatic bones, with a steep mandibular plane angle.(**Figure 2B**) The posteroanterior skull view revealed hypoplasia of the mandible and malar region, along with hypertrophy of the walls of both maxillary sinuses. (**Figure 2C**)

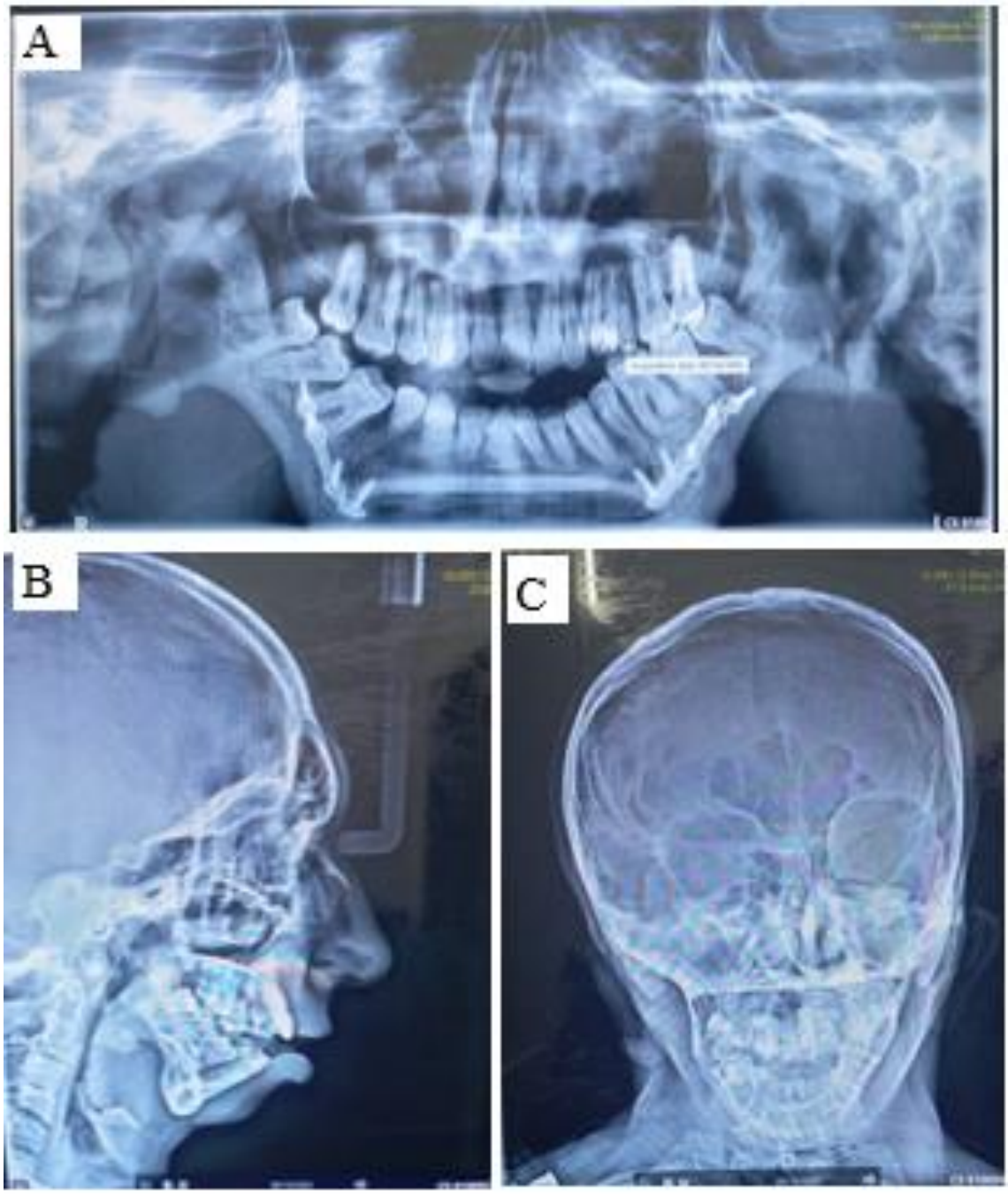

Figure 2: Radiographic presentation of patient with Treacher Collin Syndrome; **A:** Orthopantomograph; **B:** Lateral cephalogram; and **C:** PA view.

3. Discussion

Treacher Collins Syndrome is a congenital disorder that affects the development of facial bones and soft tissues. The severity of the condition varies widely among individuals. TCS influences the size, shape, and position of the jaws, cheekbones, eyes, ears, and eyelids. During embryonic development, several genes play crucial roles in craniofacial morphogenesis, and mutations in any of these genes can result in the syndrome. Although additional genetic factors may still be unidentified, mutations in three genes such as TCOF1, POLR1C, and POLR1D have been strongly associated with TCS. In approximately half of the cases, the syndrome is inherited in an autosomal dominant pattern, whereas in the remaining cases, it results from de novo mutations without a family history. More than 60% of individuals diagnosed with TCS have no familial background of the disorder. The absence of a positive family history in the present case suggests a sporadic mutation rather than inherited transmission of the TCOF1 gene.[6]

TCS primarily affects the craniofacial complex, which includes the skull, eyes, ears, nasal structures, facial bones, and orodental tissues. The clinical presentation is typically symmetrical and evident at birth; however, in the present case, the facial hypoplasia was more pronounced on the left side despite being bilateral.

3.1. Clinical features of TCS

3.1.1. Ophthalmic features

Key ocular manifestations include notching of the eyelids (coloboma), underdeveloped lower eyelids and lateral canthi, and sparse or missing eyelashes, particularly medially. Downward slanting of the palpebral fissures is characteristic. Less common findings include amblyopia, anisometropia, strabismus, hypertelorism, congenital cataracts, or microphthalmia. Tear drainage anomalies such as dacryostenosis may also occur.

3.1.2. Orbital abnormalities

The orbital bones often exhibit hypoplasia, especially along their lateral walls.

3.1.3. Otological abnormalities

External ear deformities, including microtia, malformed or low-set ears, and narrowing or absence of the external auditory canal (canal atresia), are common. Middle ear malformations may accompany these findings, frequently resulting in conductive hearing loss of up to 60 decibels.

3.1.4. Facial bone malformations

A hallmark feature is a bilaterally symmetrical, convex facial profile due to hypoplasia of the zygomatic arches and malar bones.

3.1.5. Maxillary and mandibular deformities

Both the maxilla and mandible typically exhibit underdevelopment, with the mandible positioned posteriorly. This retrognathia may lead to temporomandibular joint dysfunction and a steep mandibular plane angle.

3.1.6. Nasal and oral manifestations

Respiratory difficulties can result from micrognathia, maxillary hypoplasia, and posterior displacement of the tongue, which may obstruct the airway. A high-arched or cleft palate (with or without cleft lip), microstomia, and a prominent nose with associated structural deformities are also observed.

3.1.7. Dental abnormalities

Dental findings may include malocclusion, hypodontia, enamel hypoplasia or opacities, spacing between teeth, skeletal open bite, and abnormal eruption patterns of the maxillary molars, contributing to the characteristic “bird-like” facial appearance.

3.1.8. Other minor manifestations

Additional features can include preauricular hair displacement, airway irregularities, nasal passage obstruction, tracheostomy presence, and systemic anomalies such as congenital heart defects, undescended testes, limb deformities, renal abnormalities, and absence of parotid glands. Developmental delays in speech or motor skills have also been reported.[8]

3.2. Differential diagnosis

The differential diagnosis for TCS includes Nager’s syndrome, Miller acrofacial dysostosis, oculoauriculovertebral spectrum, Goldenhar syndrome, CHARGE syndrome, and Pierre Robin sequence.[9]

3.3. Treatment plan and management

The management of TCS requires a multidisciplinary approach involving oral and maxillofacial surgeons, orthodontists, otolaryngologists, ophthalmologists, and speech therapists. The treatment strategy focuses on functional rehabilitation, aesthetic correction, and psychosocial support, tailored to the patient’s age, severity of deformity, and individual needs.

1. *Airway obstruction*
   In neonates with airway obstruction, mandibular distraction osteogenesis can be performed to advance the mandible and improve airway patency. If the airway remains stable, the procedure may be deferred until early childhood.
2. *Mandibular retrognathia*
   In patients presenting with mandibular deficiency, mandibular lengthening is typically undertaken between 2–3 years of age or later. Reconstruction with

autogenous bone grafts is generally completed before addressing associated soft tissue defects. Prior to the completion of facial growth, procedures such as chin augmentation or mandibular distraction may be performed to improve facial balance. Definitive orthognathic surgeries, including bilateral sagittal split osteotomy, segmental maxillary surgery, or Le Fort I osteotomy, are usually planned after skeletal maturity (around 16–18 years of age).

3. *Nasal, ocular, and ear corrections (Late Childhood)*
   - A high nasal bridge can be corrected using conventional rhinoplasty techniques.
   - Eyelid coloboma is managed through Z-plasty procedures.
   - Ear reconstruction may be achieved using prosthetic auricular replacements, and hearing rehabilitation can be supported with a bone-anchored hearing aid (BAHA) when indicated.
4. *Cleft palate*

   Palatoplasty is ideally performed within the first year of life to restore palatal function and facilitate normal speech development. Postoperatively, patients should undergo regular evaluation by an otorhinolaryngologist, followed by speech therapy to enhance communication skills.
5. *Dental and orthodontic care*

   Given the complex craniofacial anomalies in TCS, continuous dental monitoring and periodic reassessment are essential throughout growth and development.

   a. *Orthodontic management*
   - Correct malocclusion (commonly Class III).
   - Align teeth to improve function and aesthetics.
   - Prepare dental arches for potential surgical interventions such as orthognathic surgery or implant placement.
   - Enhance facial profile and occlusal harmony.
   - Facilitate improved mastication, speech, and oral hygiene.

   b. *Periodontal management*
   - Assess gingival and alveolar bone health regularly.
   - Detect early signs of periodontal disease, particularly in regions affected by crowding or malocclusion.
   - Evaluate the influence of craniofacial deformities on oral hygiene maintenance.
   - Establish a baseline periodontal status before initiating orthodontic or surgical treatment.
   - Implement preventive measures to maintain periodontal stability throughout the treatment process.

4. Conclusion

Each patient with Treacher Collins Syndrome presents with a unique pattern of clinical manifestations and severity. Many of the craniofacial deformities associated with the condition can be effectively managed through appropriately planned surgical interventions. Comprehensive treatment planning, supported by thorough clinical and radiographic evaluation as well as detailed medical and family history, is essential for optimal outcomes. Equally important is addressing the psychosocial well-being of both the patient and their family, ensuring holistic care and long-term rehabilitation.[6]

5. Source of Funding

None.

6. Conflict of Interest

None.

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