

## A Rare Case of Treacher Collins Syndrome

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**Abstract:** Treacher Collins syndrome (TCS) (mandibulofacialdysostosis or zygoauromandibular dysplasia) is one of a group of congenital malformation syndromes that have in common maldevelopment of the first and second branchial arches. Treacher Collins syndrome is an autosomal dominant disorder of craniofacial development which has an incidence of approximately one in 50,000 live births. Here we report the case of a 49-year-old male with TCS

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### I. Introduction

Treacher Collins syndrome is a severe congenital disorder of craniofacial development characterized by numerous developmental anomalies that are restricted to the head and face. Hypoplasia of the facial bones, particularly the mandible and zygomatic complex, is an extremely common feature of TCS. Although the condition was probably first described by Thomson (1846) followed by Toynbee (1847) and Berry (1889), it is named after E Treacher Collins who described the essential components of the syndrome in 1900. The first extensive review of the condition was done by Franceschetti and Klein in 1949, who coined the term mandibulofacialdysostosis to describe the clinical features. In England and on the American continent this abnormality is described as the “Treacher Collins syndrome” and on the European Continent as “Mandibulofacial dysostosis” or “Franceschetti syndrome”

### II. Case Report

A 49 years old male admitted in medicine ward with complains of recurrent cough and cold with nasal congestion. The patient is the second child of the parents, who was born at term by a normal vaginal delivery at home conducted by dai. No history of neonatal ICU admission or hospitalisation in the past. He was born of a non-consanguineous marriage and the pedigree analysis reveals that no other family member was previously affected in the family. The patient was immunized up-to-date. The developmental milestones were achieved on time except for a hearing deficit. On general examination the weight was 45 kg and had an average built. The facial characteristics revealed facial dysmorphic features. The nose was well-developed with broad base leading to hypertelorism. A downward slanting of eyes (anti-mongoloid), malar hypoplasia, micrognathia.

The pinna of both ears had an abnormal appearance. The size of both the ears was small (microtia) and external auditory canal atresia was present in both the ears. Oropharyngeal examination revealed a high arched palate and bilateral tonsillar hypertrophy. No anomaly of limbs was observed. The visual acuity in the right eyes was 6/6 and in the left eye was 6/9 improving to 6/6 with pinhole. The Lower lid coloboma was observed in right lower eyelid at the junction of medial 2/3<sup>rd</sup> and lateral 1/3<sup>rd</sup>. Mild ptosis was observed in the left eye with the upper lid covering 1/3<sup>rd</sup> of the cornea. Pupils of both eyes were normal size reacting to light. The fundus examination of both eyes was normal.



Hypoplasia of the zygomatic complex and eyelashes absent in medial two-thirds of the lower eyelid of both eyes

### III. Discussion

As mentioned earlier Treacher Collins Syndrome (TCS) is an autosomal dominant hereditary disorder, hence genetic factor plays a major role in its etiology.

TCS is caused by mutation of the *TCOF1*, *POLR1C* or *POLR1D* genes. In the case of *TCOF1* the mode of inheritance is autosomal dominant, while for *POLR1C* it is autosomal recessive and for *POLR1D*, it can be autosomal dominant or autosomal recessive.

Treacher Collins presents itself with various clinical presentations and varies in affected individuals, even if among family members (Rovin et al., 1954). The syndrome commonly affects the bones of the face, ears and orbit, and usually have a bilateral presentation. Respiratory complications may sometimes be fatal and the clinical presentation may be complicated due to the fact that respiratory ventilation is difficult to achieve due to defective facial bone development (Negamine and Kurahashi, 2007; Jayasekera, 2007). TCS presents either with ears that appear normal, or the external ears may be abnormally small or completely absent. Atresia or stenosis of the external auditory canals, conductive hearing loss (ranging from mild to severe) usually due to malformations of structures within the middle ear (Herberts, 1962). Our case also showed deformity of the left ear with absence of left auditory canal with associated conductive hearing loss too. Eye abnormalities can give a saddened facial appearance to the affected individuals. Lower eyelid abnormalities increases the risk of eye infection. Specific ocular eye defect associated includes coloboma, drooping eyelids, downward slanting palpebral fissures, partial or complete absence of the lower eyelashes, dacryostenosis or narrowed tear ducts, also vision loss (Roy, 1978; McEnery & Brennemann, 1937; Poswillo, 1976). Hypoplasia of the malar bones and mandible are classic facial features of TCS which complied in our case too. The retrognathic mandible gave a relatively prognathic anterior maxilla which indirectly led to incompetent lips. Approximately 55 to 60% of individuals with TCS may develop dental abnormalities including tooth agenesis, widely-spaced teeth, malocclusion, enamel opacities, ectopic eruption of maxillary first molars, macrostomia. Other findings, which less frequently occur are: High arched palate, nasal deformity, cleft palate (McEnery & Brennemann, 1937; Poswillo, 1976). Our case had enamel hypoplasia, high arched palate with cleft and deformed bifid uvula. In the differential diagnosis, one should consider the acrofacial dysostoses. Acrofacial dysostoses such as Nager syndrome and Miller syndrome have appearances that resembles that of Treacher Collins syndrome, but additional limb abnormalities occur in those patients. Hemifacial microsomia which primarily affects development of the ear, mouth, and mandible can be considered a differential diagnosis, however this anomaly may occur bilaterally. Another disease which belongs to this spectrum is Goldenhar syndrome, which includes vertebral abnormalities, epibulbar dermoids and facial deformities. The classic ocular presentation in Goldenhar syndrome, ruled out this differential diagnosis hence confirming the initial diagnosis of Treacher Collins Syndrome.

### IV. Conclusion

Conclusion From the above discussion it is very clear that Treacher Collins syndrome is a rare manifestation and needs more focus on, improving the diagnostic ailments, as the critical period of pathogenic activity for deformities occurs around 7 weeks in utero, and to improve the current approach in treatment modalities from aesthetical correction to psychosocial development and also to bring in the multidisciplinary team of plastic surgeons, maxillofacial surgeons, otorhinolaryngologist, ophthalmologist, speech therapist and psychologist in order to offer better quality of life to patients as well as their families.

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