CASE REPORT

Treacher Collins syndrome - A case report

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Abstract

Treacher Collins syndrome (TCS) is a genetic disorder that arises during early embryogenesis and is characterized by abnormality of craniofacial development. It is generally characterized by bilateral symmetrical abnormalities of structures which fall within the first and the second branchial arches. The main clinical findings include midface hypoplasia, micrognathia, microtia, conductive hearing loss, and cleft palate. In this article, a case report of 40-year-old male presenting with TCS is discussed briefly.

Keywords: Autosomal trait, mandibulofacial dysostosis, Treacher Collins-Franceschetti syndrome

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Introduction

Treacher Collins syndrome (TCS) is a rare autosomal dominant disorder of craniofacial development known for its high penetrance and variable expressivity. It roughly affects 1/50,000 live births.[1] Although TCS was first described in 1846 and 1847 by Thomson and Toynbee, respectively, and by Berry in 1889, it is named after E Treacher Collins, a British ophthalmologist who reported 2 cases of TCS in 1900. Franceschetti and Klein,[2] in 1944, described the cases with an extensive review, thereby proposing the term “mandibulofacial dysostosis.” The etiology of the syndrome is attributed to the mutations of genes TCOF1 (5q32-q33.1) which encodes for nucleolar phosphoprotein Treacle or the POLR1C gene (6p21.1) and POLR1D gene (13q12.2), encoding for RNA polymerases I and III subunits.[3]

Symptoms of this syndrome vary widely, which ranges from almost unnoticeable to mild to severe. Classic findings in mandibulofacial dysostosis include antimongoloid slanting of the palpebral fissures, temporal lower eyelid coloboma, malar and mandibular hypoplasia, malformation of the auricle associated with atresia of the external auditory canals and maldevelopment of middle ear ossicles, and associated conductive hearing loss.

Case Report

A 40-year-old male patient reported to the institute complaining of difficulty in chewing food due to missing teeth and wanted to replace the same with an artificial denture. The patient has been partially edentulous since childhood and started using dentures later. He gave history that some of his teeth were partially erupted during childhood, which were yellowish in color and exfoliated within a short period of time. On general examination, the patient was of short stature and low weight. Extraoral findings revealed long narrow face with hypoplasia of malar bones bilaterally, antimongoloid slant of both palpebral fissures, inferior palpebral coloboma, abnormality of external ear, absence of pinnae, abnormal hair growth pattern extending from hairline toward the cheek in the temporal region, and a prominent nose over a hypoplastic maxilla, but no significant reduction in neither hearing nor retrusion of chin was seen [Figures 1-2]. Family history was negative for related features and patient demonstrated an age-appropriate mental and speech development. Intraoral examination revealed a narrow high-arched palate and edentulous maxillary and mandibular alveolar ridges [Figures 3 and 4]. On radiographic examination, we found hypoplasia of the mandible and maxillary sinus, short ramus, and prominent antegonial notch [Figures 5 and 6]. Based
on clinical features and radiographic findings, diagnosis of TCS was reached. Our treatment plan included necessary complete dentures after appropriate evaluation of the alveolar ridges in the department of prosthodontics.
Discussion

Treacher Collins syndrome (TCS) is a condition of craniofacial development associated with several head and neck defects without abnormalities of the extremities. The cause of autosomal dominant TCS is suggested to be heterozygous mutation in TCOF1 or POLR1D gene, whereas syndrome with mutations in POLR1C is inherited in an autosomal recessive manner. Two-thirds of cases are known to arise de novo without any previous family history; the remaining are known to inherit one mutated copy of the TCOF1 gene from either parent which will ultimately lead to haploinsufficiency.\

The classic features of TCS are evident at birth and also they are bilaterally symmetrical. The diagnosis becomes easy on clinical characteristics alone only when there is full expression of the syndrome. Thus, it is easy to diagnose mild cases, only by clinical examination but difficult to diagnose severe cases as it results in profound developmental abnormalities, leading to perinatal mortality. Although TCS shows high penetrance of the genetic mutations, significant inter- and intra-familial clinical variability is a remarkable feature and reflects a multifactorial basis. The pathogenesis of TCS was explained by various hypotheses which included abnormal patterns of neural crest cell migration, improper cellular differentiation during development, abnormal domains of cell death, or an abnormality of the extracellular matrix. In TCS patients, the haploinsufficiency of treacle might cause insufficient rRNA production in the prefusion neural folds which results in craniofacial developmental abnormality. The survival and proper differentiation of cephalic neural crest cells during early embryogenesis probably require a higher threshold concentration of rRNA as suggested by Valdez et al.\

Franceschetti and Klein described the typical characteristics of the syndrome, later Axelsson et al. (1963) specified the obligatory features in which he included antimongoloid palpebral fissures, coloboma of the outer third of the lower eyelid, or deficient lashes, or both, as well as the hypoplasia of the malar bones and mandible.\

Indeed, some patients are so minimally affected that it is difficult to reach a diagnosis. Hence, minimal diagnostic criteria such as hypoplasia of zygomatic arch and downward slanting palpebral fissures for the diagnosis of TCS were proposed by Teber et al.\

Five clinical forms of TCS were described by Franceschetti and Klein as the complete form, incomplete form, the abortive form, unilateral form, and the atypical form.\

In our case, the patient presented incomplete form of the syndrome.\n
Dental anomalies were found by da Silva Dalben et al. in 60% of individuals with TCS. The identified anomalies were enamel opacities, tooth agenesis, and ectopic eruption of the maxillary first molars.\n
Characteristic radiographic findings of TCS include hypoplasia or aplasia of the zygomatic arch, malar hypoplasia, mandibular retrognathia, irregular or absent auditory ossicles, complete absence of the middle ear and epitympanic space, and partial absence of the stapes.\n
Thus, the diagnosis of this syndrome is based on clinical features and complementary examinations and supported by molecular tests to confirm the diagnosis. Treatment should be of multidisciplinary in approach where craniofacial management team should plan and tailor to the specific needs of each individual.\n
Conclusion

TCS is a rare genetic disorder, the early diagnosis and appropriate treatment of which will help in improving the esthetic and functional deficiencies in these patients. Management is multidisciplinary and early intervention will help to obtain better therapeutic results.

References
