Treacher Collins Syndrome

\textsuperscript{1}Alexander, \textsuperscript{2}Peter Sherry

\textsuperscript{1}Professor, Department of Oral Medicine and Radiology, Amrita School of Dentistry, Cochin, Kerala, India
\textsuperscript{2}Consultant, Department of Head and Neck Surgery, Amrita Institute of Medical Sciences, Cochin, Kerala, India

Correspondence: Alexander, Professor, Department of Oral Medicine and Radiology, Amrita School of Dentistry, House No. 13, Surya Gardens, Marottichode, Edapally, Cochin, Kerala-682024, India, e-mail: alexvinod@gmail.com malexander@aims.amrita.edu

Abstract
Treacher Collins syndrome is a rare autosomal dominant condition, predominantly affecting the orofacial structures. The incidence varies between 1 in 40,000 to 1 in 70,000 per live births. 40\% of the cases have a hereditary factor while 60\% are due to genetic mutations. The features include antimongloid slanting of the eyes, deformed or underdeveloped pinna of the ear, retrognathic mandible, microgenia, hypoplasia of the facial bones. In some patients, the retrognathic mandible may cause difficulty in breathing and swallowing and may require surgical interventions. The present article describes the clinical features of Treacher Collins syndrome as seen in 3 cases.

Keywords: Mandibulofacial dysostosis, Treacher Collins syndrome.

INTRODUCTION

Treacher Collins Syndrome (TCS) is an autosomal dominant condition with varying degree of penetrance and expression. It affects mainly the head and face.\textsuperscript{1} The first description of the condition was made by Thomson in 1846\textsuperscript{2} followed by Toynebee in 1847\textsuperscript{3} and Berry in 1889.\textsuperscript{4} Treacher Collin described the condition in 1900 in two cases of lower lid coloboma and noted the deformities of the zygoma and maxilla. The condition is thus credited to his name.\textsuperscript{5} It was later further described and classified by Franceschetti et al and renamed as ‘mandibulofacial dysostosis.’\textsuperscript{6}

The most common manifestations of TCS are the antimongloid slanting of the palpebral fissures, colobomas of the lower eyelid, hypoplastic zygoma, hypoplastic maxilla and mandible, malformations of the auricular pinna and conductive deafness. Cleft palate may or may not be associated with the syndrome.\textsuperscript{7}

This article reports three cases which show the common clinical features of TCS.

CASE REPORT

Case One (Fig. 1)

A 4-year-old female patient reported to the department of pediatrics with her parents for an evaluation of the cardiac problem. The patient was born with multiple congenital anomalies—microcephaly, facial dysmorphism, malformed ears. The child was suspected of congenital heart defect 21 days after birth during evaluation for noisy breathing. The evaluation then revealed ventricular and auricular septal defects. Family history revealed the child as the first born of nonconsanguineous parents. She was delivered as a full term baby through normal delivery with a birth weight of 2.8 kg. The developmental history showed signs of poor growth and development. The developmental milestones were delayed with the child currently being unable to walk without support and unable to speak monosyllabic words.

Previous medical history revealed that she had severe airway compromise at the age of three months for which medical advice was sought, mandibular distraction was planned as the treatment modality but the patient did not turn up for the treatment on the specified day. The patient presently complained of sleep apnea due to falling back of the tongue.

General physical examination of the child revealed severe failure to thrive, severe microtia (low set deformed pinnae with anostiae), total absence of the ear canal, hypoplastic ala nasi, and severe micrognathia. Fusion of the lateral portion of the eyelids was seen bilaterally. Downward slant of the eyes were also seen. Other congenital malformations included small head, hypoplastic mandible, antimongloid slant and partially open anterior fontanelle.

Genetic evaluation of the patient revealed total atresia of ears, hypoplastic ala nasi and small palpebral fissures with fusion at the lateral portion. In addition, hypoplastic mandible,
hypoplastic zygoma and severe micrognathia were also seen. These features were in accordance with the phenotypic features of TCS.

Oral examination of the patient revealed difficulty to open the mouth, hypoplastic mandible and presence of complete deciduous dentition. Anterior open bite was also seen. The patient did not show or complains of any signs of air compromise, but medical history revealed that difficulty in breathing was present during upper respiratory tract infection.

**Case Two**

A 15-year-old male patient reported to the head and neck institute with a chief complaint of facial deformity. The patient was born with hypoplasia of the mandible and zygomatic bone and other congenital malformations. Family history revealed the child as the first born of nonconsanguineous parents. There was no similar deformity in the siblings of the patient. On physical examination, he presented with antimongloid slanting of the palpebral fissures with sparse eyelashes on the lower eyelid (Fig. 2). There was hypoplasia of the malar prominence. There was absence of both auricles bilaterally except for the presence of the lobules. Microgenia was also evident (Fig. 3). The maxilla appeared to be prognathic. On examination of the feet and hands, metatarsus adductus deformity was seen on the feet.

Intraoral examination revealed narrow high arched palate. The maxillary central incisors were overlapped and there was mild crowding in the mandibular anteriors. Anterior open bite was present. There were root stumps in place of 16, 26 and 27. Macroglossia was also evident. There was no evidence of airway obstruction.

Radiographic features revealed short ramus with anterior open bite (Fig. 4).

**Case Three**

A three and a half years old female patient reported to the head and neck institute with a chief complaint of decayed tooth with tenderness on right upper jaw. The child was reported to have been born with facial deformities which have been worsening as the child grows. Consanguinity was denied by the parents. On extraoral examination, bilateral malar hypoplasia, presence of coloboma, presence of antimongolid slant, absence of medial eyelashes (Fig. 5), bilateral deformed pinnae were seen. Severe mandibular hypoplasia and anterior open bite were also present (Fig. 6). It was noticed that the mouth was open and snoring was present while sleeping. Presence of obstructive sleep apnea syndrome was confirmed but not very severe.

**DISCUSSION**

Treacher Collins syndrome or Franceschetti syndrome is a rare congenital disorder. It occurs about 1 in 50,000 live births. The syndrome follows an autosomal dominant trait, affecting both genders equally. 40% of the cases reported have a familial history while 60% are new mutations and these seem to be associated with increased paternal age. The syndrome is
characterized by abnormalities which are bilaterally symmetrical. The malformations were initially thought to be due to the defective ossification of the facial bones, but as the tissues affected were derivatives of the first and second brachial arches, it was suggested that defects in the migration of the neural crest cells could be responsible. Other hypotheses for the pathogenesis of TCS were premature cell death in the ectodermal placodes of the first and second brachial arches, improper differentiation of embryonic tissues or an abnormality in the extracellular matrix. Recent evidence suggests that mutations in the gene TCOF1 or 'treacle' may be the cause of the craniofacial malformations seen in Treacher Collins syndrome. The clinical features of Treacher Collins syndrome include a typical “fish like” facial appearance, micrognathia, microgenia, macrostomia, cleft palate, coloboma of the lower eyelids, antimongloid slanting of the palpebral fissures, absence of eyelashes over the lower lid, malformation of auricles and ‘fish like’ or ‘bird like’ appearance of the face. Anterior open bite was also seen in all of the above cases. The severity of expression was varied in all the above cases. None of the above cases showed a hereditary factor suggesting that all the cases were due to new mutations affecting the TCOF1 or ‘treacle’. Sleep apnea was present in case one and case three but not severe enough to seek for treatment. Rarely congenital heart defects can be present in patients with TCS as in seen in case one.

CONCLUSION

Treacher Collin syndrome is an autosomal dominant syndrome which may present in a variety of clinical features. Management of TCS is presently limited to relief of airway obstruction in case of sleep apnea or choanal atresia. Management of TCS should be a multidisciplinary approach and enable the patient to attain better functional efficiency as well as esthetics.

REFERENCES

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