CASE REPORT

Treacher Collins syndrome: a case report

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SUMMARY

Treacher Collins syndrome is a rare autosomal dominant disorder of craniofacial development. The fully expressed phenotype exhibits characteristic dysmorphic features involving the face, eyes, mandible and ears. We report a case of a 17-year-old woman presenting with the typical orofacial implications of this syndrome.

BACKGROUND

Mandibulofacial dysostosis (MFD), or Treacher Collins syndrome (TCS), is an autosomal dominantly inherited disorder that arises from aberrations in the development of facial structures derived from the first and second branchial arches during histodifferentiation morphogenesis between approximately the 20th day and the 12th week of intrauterine life.¹ This syndrome was described by Thomson (1846), Berry (1889), Treacher Collins (1900) and Franceschetti and Klein (1949).²

Downward slanting palpebral fissures and hypoplasia of the zygomatic arch have been defined as the minimum diagnostic criteria by Teber *et al.* The present case report illustrates the orofacial features of this syndrome.

CASE PRESENTATION

A 17-year-old woman reported to our department with a sensitivity to hot and cold in her left lower back tooth. An extraoral examination revealed a narrow face with mandibular and zygomatic hypoplasia along with antimongoloid slant of eyes (figure 1). Mandibular hypoplasia caused the upper dentition to appear protruded. Malar hypoplasia resulted in a 'sunk-in' appearance temporally, causing the nose to appear very prominent. The patient's eyes were remarkable and there was partial absence of lower eyelashes and coloboma of lower lateral eyelid (figure 2). External ear malformation in the form of a rudimentary pinna was present bilaterally. In addition, the external ear canals were atresic with absence of opening from the external to the internal ear. Conduction deafness with 50% reduction in hearing was also present. Another interesting feature was the presence of a tongue-shaped process of hair on the lateral side of the face (figure 3). Intraoral examination revealed an anterior open bite and carious left lower first and second molars. This patient was the youngest of the four children born to parents with no history of consanguineous marriage. Her mother was 36 years and her father was 44-year-old at the time of her birth. None of the siblings showed these facial features. The radiographic investigations (posteroanterior (PA) skull view and lateral skull view) revealed mandibular hypoplasia along with prominent antgonial notch.

Figure 1 Profile photograph of the patient demonstrated mandibular and zygomatic hypoplasia and antimongoloid slant of eyes.

Overall, clinical and radiographic features were suggestive of TCS.

INVESTIGATIONS**Radiographs:**

- ▶ Extraoral PA skull view demonstrated malar and mandibular hypoplasia along with hypertrophy of both maxillary sinus walls (figure 4).
- ▶ Lateral skull revealed prominent antgonial notch and anterior open bite (figure 5).

Figure 2 Partial absence of lower eyelashes and coloboma of the lower lateral eyelid.

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Figure 3 External ear abnormality and tongue-shaped process of hair on lateral aspect of face.

Figure 4 Posteroanterior skull showing mandibular and malar hypoplasia with hypertrophy of both maxillary sinus walls.

Figure 5 Lateral skull revealed prominent antegonial notch and anterior open bite.

DIFFERENTIAL DIAGNOSIS

- ▶ Nager's acrofacial dysostosis
- ▶ Milleracrofacial dysostosis
- ▶ Oculoauriculovertebral spectrum
- ▶ Goldenhar syndrome

TREATMENT

The patient was treated for her chief complaint and was motivated for orthodontic treatment and prosthetic reconstruction of the ears.

OUTCOME AND FOLLOW-UP

The prognosis for the current case was fair.

DISCUSSION

TCS, alternatively called MFD, is an autosomal craniofacial development disorder related to the chromosomal region 5q32–q33.1, presenting peculiar facial aspect and clinical and genetic heterogeneity.³ A total of 51 mutations in the TCOF1 gene had been identified to date, all of which result in introduction of premature termination codons into the reading frame, suggesting haploinsufficiency as the molecular mechanism underlying the disorder.⁴ The incidence is estimated at 1 in 50 000 live births; nearly 40% of cases present familial history, whereas the remaining 60% are considered new mutations. Its occurrence is influenced by the increase in paternal age.³ The adult patient with fully expressed TCS has a convex facial profile with a prominent dorsum of the nose above a retrusive lower jaw and chin. The eyes are characterised by an antimongoloid slant of the palpebral fissure resulting from coloboma and hypoplasia of the lower eyelids and lateral canthi, including partial absence of eyelid cilia.^{3 5} 'Tongue-shaped' processes of hair frequently extending into the pre-auricular region. The external ears are

absent, malformed or malposed, and hearing was impaired as a result of variable degrees of hypoplasia of the external auditory canals and ossicles of the middle ears.⁵ All the above-mentioned features were present in our case. Macrostomia or alternatively microstomia with a narrow and high palatal vault, and a cleft palate in about 35% of cases is also seen.⁶ Palate deformities were not present in our patient; however, intraorally, anterior open bite was present. Dental misalignments with anterior open bite are frequent in this syndrome.⁶ Other dental anomalies include supernumerary teeth, T-shaped teeth, enamel opacity, enamel hypoplasia, tooth agenesis, microdontia, tooth rotations and ectopic tooth positioning.³ The characteristic facies in TCS occur as a result of the destruction of the neural crest cells before they migrate to form the facial processes. Normally derived structures of the first and second branchial arches exhibit malformation.¹ A diagnosis was made based upon a thorough clinical evaluation, detailed patient history and identification of characteristic findings. Specialised imaging techniques such as x-ray or CT may be performed to assess the extent of certain craniofacial abnormalities such as middle and inner ear structures.⁷ Molecular genetic testing to confirm a diagnosis is available through commercial and academic research laboratories to detect mutations in the TCOF1, POLR1C and POLR1D genes. Approximately 90–95% of individuals have an identifiable mutation of the TCOF1 gene. Prenatal screening via ultrasound during mid-to-late gestation may detect cases with severe craniofacial abnormalities. Relatives, especially parents and siblings, of an individual diagnosed with TCS should be carefully examined because mild cases often go unnoticed and undiagnosed.⁷ Treatment of MFD (TCS) is lengthy and requires a multidisciplinary approach focused on treatment of symptoms. In newborns with MFD, immediate attention to airway and swallowing inadequacies is critical. A tracheostomy (a hole into the trachea through the front of the neck) may be needed to help breathing, while reconstructive surgery may be necessary to correct a cleft palate. A notch, or coloboma, in the lower eyelid may cause the eyes to dry out easily, increasing the risk of infection. This may require surgery. Reconstructive surgery may also be offered to build up the cheekbones, correct the nose or an underdeveloped jaw (which may help to improve breathing) or reconstruct the outer ear.^{7–9} Other treatment includes distraction osteogenesis associated with preoperative and post-operative orthodontic treatment leading to a better quality of life.⁸ Surgical reconstruction of the auricle can be satisfactorily achieved through a staged approach in the hands of a few experts. The successful grafting of a well-sculpted cartilage framework is the foundation for a sound auricle repair. Other stages of the auricular construction include lobule transposition, detaching the auricle with a skin graft, managing the hairline and reconstructing the tragus.⁵ TCS is an example of an autosomal dominant syndrome with incomplete penetrance and variable expressivity. An affected parent of either sex will transmit

the defect to 50% of offsprings in accordance with Mendelian laws of genetics. This emphasises the importance of genetic counselling to affected individuals. It is our responsibility as oral physicians to recognise this disorder, to be aware of its manifestations and to provide close follow-up, appropriate therapy and counselling.⁹ Also, early diagnosis of TCS allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients. In fact, ameliorating the outward signs gives these patients the opportunity to have an improved social life.⁸

Learning points

- ▶ Early diagnosis of Treacher Collins syndrome allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients. If this can be performed early, it is possible to take advantage of anticipated growth during normal skeletal maturation and to obtain better therapeutic results.
- ▶ In severe cases the airway must be evaluated and secured from birth. Either positioning alone or tracheostomy is required to manage the airway, and a gastrostomy required for feeding.
- ▶ Genetic counselling can help families understand the condition and how to care for the patients.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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