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OLGU SUNUMU / CASE REPORT

Treacher Collins syndrome: a case report

Treacher Collins sendromu: bir olgu sunumu

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Abstract
Treacher Collins Syndrome or mandibulofacial dysostosis is a rare congenital malformation involving first and second branchial arches and presents several craniofacial deformities. The occurrence of this syndrome is estimated to range between 1 in 40,000 to 1 in 70,000 live births. The syndrome is characterized by anomalies of the auricular pinna, hypoplasia of facial bones, antimongoloid slanting palpebral fissures, coloboma of the lower eyelids and cleft palate. Here we report a case of Treacher Collins Syndrome with a narrative review of the clinical features, radiographic findings, differential diagnosis and various treatment options.
Key words: Treacher Collins Syndrome, mandibulofacial dysostosis, congenital malformation

Öz
Treacher Collins Sendromu veya mandibulofasiyal disostosis çeşitli kraniyofasiyal deformasyonlar olarak ortaya çıkan birinci ve ikinci brankial kemerleri içeren nadir bir konjenital bozukluktur. Bu sendromun oluşmasının 70,000 canlı doğumda 1 40.000 ile 1 arasında olduğu tahmin edilmektedir. Bu sendrom auriküler pinna anomalileri, yüz kemikleri hipoplazisi, antimonoloid sarkık palpebral çatlaklar, alt göz kapaklarının kolobom ve yarık damak anomalileri ile karakterizedir. Burada klinik özellikleri, radyografik bulgular, ayırıcı tanı ve çeşitli tedavi seçenekleri ile ayrıntılı bir inceleme ile Treacher Collins sendromu olgusu sunulmuştur.
Anahtar kelimeler: Treacher Collins Sendromu, mandibulofasyal disostoz, konjenital malformasyon

INTRODUCTION

Treacher Collins syndrome (TCS) or mandibulofacial dysostosis is an autosomal dominant disorder. The first extensive explanation of the syndrome was given by Franceschetti and Klein in 1949, who used the term mandibulofacial dysostosis. However, the essential features of this syndrome were already described by Treacher Collins in the year 1900. 60% of the autosomal dominant cases arise as de novo mutation[1,2]. It is typically characterized by abnormalities in the first and the second branchial arches, with no racial or gender predilection. TCS is related to mutations in either of the 3 genes: TCOF1, POLR1C or POLR1D[3]. Common manifestations of TCS are mandibular hypoplasia, hypoplasia of facial bones, zygoma and maxilla, anti-mongloid slanting of palpebral tissues, cleft palate, malformation of the external ear with conductive hearing loss and coloboma of the lower eyelid. Craniofacial anomalies can lead to airway obstruction and obstruction sleep apnea syndrome. The condition has variable expressivity. The severity of the clinical features often tends to be greater in subsequent generations of the same family[3].

Here we report a 20 year old female patient with the features of Treacher Collins syndrome having normal cognitive abilities and who is the only member of the entire family who is diagnosed with these syndromic features. Besides we have also given a narrative review of the various common and uncommon features that can be seen in this syndrome and their management.

CASE

A 20 year old female patient reported to the

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Department of Oral Medicine and Radiology with a chief complaint of forwardly placed upper and lower teeth since birth.

Figure 1. Facial features of Treacher Collins syndrome

Patient gives a history of associated discomfort while closing the mouth and chewing. Patient's medical history revealed that she had visited the ENT surgeon previously for hearing deficit. She has not been treated for the same. Family history revealed that her mother had a cyst in the reproductive system during the time of pregnancy. The mother's age at the time of pregnancy was 34 years and father was 36 and she was the second child among the 2 siblings. Patient was conscious, co-operative, well oriented to time, place and person, moderately built and nourished. On Extraoral examination patient had downslanting palpebral fissures, hypoplastic supraorbital rims, hypoplastic zygomas, narrow face, depressed cheek bones, receding chin, large downturned mouth and a convex profile Figure 1.

Intraoral examination revealed high arched palate, narrow constricted maxillary arch and dental caries w.r.t 17, 27, 46 and 47 and grossly decayed 35 (Figure 2). Panoramic radiograph showed bilaterally prominent antegonial notch and short ramus of the mandible, grossly decayed 35, erupting tooth buds with respect to 18, 28, 38 and 48. An enlarged pulp chamber, apical displacement of the pulpal floor and lack of constriction at the cementoenamel junction is seen in relation to 37 and 47 suggesting Taurodontism (Mesotaurodont with respect to 37 and 47) (Figure 3). Lateral Cephalogram and Posteroanterior view of the skull reveals hypoplastic mandible, prominent nose and proclined upper anteriors, hypoplasia of zygomatic bone and maxillary sinus, prominent antegonial notch, acute nasolabial angle, underdeveloped maxillary sinuses, malar bones and hyperteloric orbits (Figure 4).

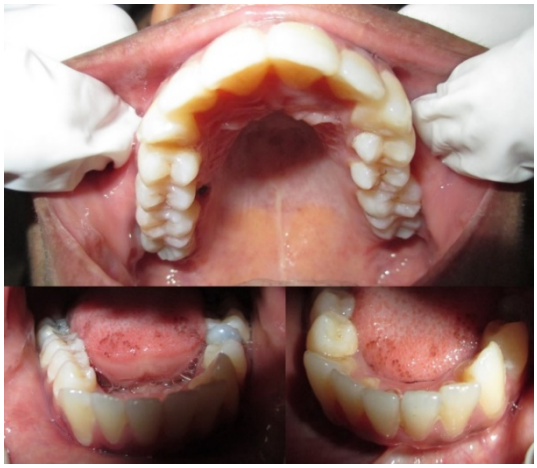

Figure 2. Intraoral photograph showing maxillary and mandibular arch.

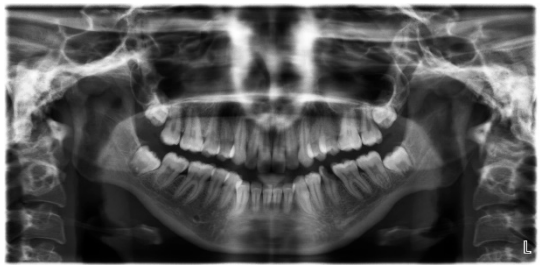

Figure 3: Panoramic radiograph showing bilaterally prominent antegonial notch, short ramus of the mandible and the presence of taurodontism

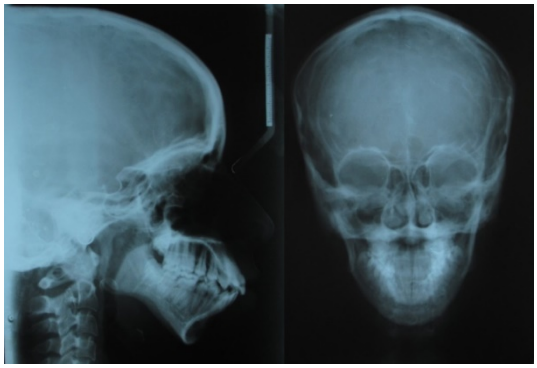

Figure 4. Lateral cephalogram and posteroanterior view of the skull revealing features of treacher collins syndrome

A provisional diagnosis of Treacher Collins syndrome was arrived at. Nager's acrofacial dysostosis, Miller acrofacial dysostosis and oculoauriculovertebral spectrum were considered as differential diagnosis.

DISCUSSION

Treacher Collins syndrome gets its name from a British Ophthalmologist Dr. E. Treacher Collins who described two children, in the year 1900, who had very small cheek bones and notches in their lower eyelids. It is an autosomal dominant disorder of craniofacial development that affects both genders equally. 40% of cases have a family history, while the remaining 60% appear to arise as a result of a de novo mutation. In our case, patient did not have a family history and hence is a de novo mutation[4].

The pathogenesis of TCS has been explained on the basis of several different hypotheses. Researchers have proposed the possibility of abnormal patterns of neural crest cell migration, abnormal domains of cell death, improper cellular differentiation during development or an abnormality of the extracellular matrix. Few researchers have come up with the possible cause to be an inhibitory process taking place towards the seventh week of the embryonic life and affecting the facial bones derived from the first visceral arch. Few others have proposed the etiology to be a defect of the stapedial artery which causes abnormal development in its own field of as well as in the region of the first visceral arch. Of late, genetic mapping procedures have recognized the mutated gene as TCOF1 and mapped to human chromosome 5q32–33.2 locus encoding a serine/alanine – rich protein called 'treacle'[5].

The derivatives first and second pharyngeal arch, groove and pouch are the ones that are affected in TCS. The characteristic features are antimongoloid palpebral fissures with either a notch or coloboma of the outer third of the lower lid, and occasional absence or paucity of the lashes of the lower lid, hypoplasia of the facial bones, especially the malar bones and mandible, malformation of the external ear, and occasionally of the middle and inner ear, with low implantation of the auricle, macrostomia, high palate, malocclusion and abnormal position of the teeth, atypical hair growth in the form of tongue-shaped processes of the hair-line extending towards the cheeks in the pre-auricular region, occasional association with other anomalies, such as obliteration of the naso-frontal angle, pits or clefts between the mouth and ear, and skeletal deformities[6].

Few syndromes have similar characteristics as that of Treacher Collins syndrome. In the differential diagnosis, one should consider the acrofacial dysostoses (Nager syndrome and Miller syndrome). The facial features resemble that of Treacher Collins syndrome, but additional limb abnormalities occur in patients with acrofacial dysostosis. The oculoauriculovertebral spectrum (Hemifacial Microsomia and Goldenhar syndrome) should also be considered in the differential diagnosis. Hemifacial macrosomia primarily affects development of the ear, mouth, and mandible. This anomaly may occur bilaterally. Goldenhar syndrome includes vertebral abnormalities, epibulbar dermoids and facial deformities[7].

Prenatal diagnosis for TCS is possible by two dimensional and three dimensional ultrasonography. It can be done if family history is positive, by doing chorionic villus sampling between 10-12 weeks of pregnancy or amniocentesis between 14- 18 weeks and can also be done by high resolution ultrasound between 18-20 weeks of pregnancy. Yet, it is difficult to diagnose mild forms of TCS with ultrasound. Postnatal diagnosis is done by assessment of clinical features and confirmation by DNA examination of affected gene[8].

Management of TCS is prolonged and requires a multidisciplinary approach that focuses on treating the symptoms. Excellent results are attainable through a comprehensive, well-coordinated and integrated treatment plans incorporating craniofacial surgeons, orthodontists, ophthalmologists, otolaryngologists and speech pathologists. A conductive hearing aid, cleft palate surgical correction, surgical repair of eye socket, correction of cheek bone, chin and eye lid notches. Skills of a peadiatric dentist is required for correction of anomalies in teeth[9].

Treacher Collins' syndrome is a rare inherited autosomal dominant condition. They need functional, aesthetical corrections, as well as psychosocial support. Multidisciplinary approach including otorhinolaryngologists, craniofacial surgeons, ophthalmologists, speech therapists, psychologists and dental surgeons is the most suitable way to manage these patients. Apart from this, such patients also carry a social stigma because

of the severe facial deformities and hence pschychiatric assistance is necessary.

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