TREACHER COLLINS SYNDROME : A CASE REPORT

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ABSTRACT

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 female presenting with the typical orofacial implications of this syndrome.

Keywords : Treacher Collins syndrome; Mandibulofacial dysostosis; Orofacial features.

INTRODUCTION

Mandibulofacial dysostosis (MFD), or Treacher-Collins syndrome, is an autosomal dominantly inherited disorder that arises from aberrations in the development of the 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), Bercy (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[1] .The present case report illustrates the orofacial features of this syndrome.

CASE REPORT

A 17 year old female reported to our department with the chief complaint of sensitivity to hot and cold in her left lower back tooth. An extraoral examination revealed a narrow face with mandibular and zygomatic hypoplasia. 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 for a downward obliquity of palpebral fissures (antimongoloid slant) and coloboma of the lateral lower eyelids (figures 1 and 2). A partial absence of lower eyelashes was also noted (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 (figure 3). 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. PA skull and lateral skull views demonstrated bilateral malar and mandibular hypoplasia along with hypertrophy of both maxillary sinus walls. A prominent antigonial notch could also be appreciated (figures 4 and 5). This patient was the youngest of the four children born to parents with no history of consanguineous marriage. Her mother was 36 years and father was 44 years old respectively at the time of her birth. None of her siblings showed these facial features. The patient was treated for her chief complaint and was motivated for orthodontic treatment and prosthetic reconstruction of ears.

DISCUSSION

The Treacher-Collins syndrome 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 Treacher Collins syndrome has a convex facial profile with a prominent dorsum of the nose above a retrusive lower jaw and chin. The eyes are characterized by an antimongoloid slant of the palpebral fissure resulting from colobomata and hypoplasia of the lower eyelids and lateral canthi, including partial absence of eyelid cilia. 'Tongue-shaped' processes of hair frequently extend into the pre-auricular region. The external ears are absent, malformed, or malposed, and hearing is 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 malalignments 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 Treacher Collins syndrome 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 second and first branchial arches exhibit malformation[1]. The diagnosis of mandibulofacial dysostosis is, in the case of full expression of the syndrome, easily made on clinical appearance[7]. Treatment includes distraction osteogenesis associated with preoperative and postoperative 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]. Treacher Collins syndrome 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 his or her offspring in accordance with mendelian laws of genetics. This emphasizes the importance of genetic counseling to affected individuals. It is our responsibility as oral physicians to recognize this disorder, to be aware of its manifestations and to provide close follow-up, appropriate therapy and counseling[9]. Also, early diagnosis of TCS allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients. Infact, ameliorating the outwards signs gives these patients the opportunity to have an improved social life[8].

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FIGURE LEGENDS

Fig. 1 Mandibular and zygomatic hypoplasia and antimongoloid slant of eyes.
Fig. 2 Partial absence of lower eyelashes and coloboma of the lower lateral eyelid.
Fig. 3 External ear abnormality and tongue shaped process of hair on lateral aspect of face.
Fig. 4 PA skull showing mandibular and malar hypoplasia with hypertrophy of both maxillary sinus walls.
Fig. 5 Lateral skull showing prominent antigonial notch and anterior open bite.