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Case Report

Radiographic Features of Treacher Collins syndrome: Case Report

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Abstract

Treacher Collins syndrome (TCS) is characterized by hearing loss, hypoplasia of the mandible, maxilla and zygoma, slanted palpebral fissures, microphthalmia of the lower lid and bilateral alteration of the external ear. In most cases, the first noticeable indicator of this disorder is facial bone hypoplasia. This article will discuss the radiographic and extraoral features in a child diagnosed with TCS who reported to the Division of Oral and Maxillofacial Radiology.

Keywords: *Treacher Collins Syndrome; CBCT; Hypoplastic Zygoma; Microphthalmia; Radiographic Interpretation*

Abbreviations

TCS: Treacher Collins Syndrome; CBCT: Cone Beam Computed Tomography; CT: Computed Tomography

Introduction

In 1900, Dr. E Treacher Collins, an English ophthalmologist, discovered tiny zygomatic bones and microphthalmia of the lower eyelid in two small children. The condition was named after this doctor [1]. Treacher Collins syndrome (TCS) is a craniofacial disorder with high penetrance and variable expressivity [2,3]. It is an autosomal dominantly inherited disorder that arises from aberrations in the development of facial structures derived from the first and second branchial arches; these aberrations manifest between approximately the 20th day and the 12th week of intrauterine life during histodifferentiation morphogenesis [4]. Its incidence is approximately 1 in 50000 live births, transmission is autosomal dominant with penetrance close to 100%, and variable expressivity is approximately 40% [5]. When affected parents with TCS give relevant genes, the children may be affected to different degrees. The degree varies and could be the same as, milder, or more severe than the parent [1].

Presentation of the Case

An 8-year-old male child diagnosed with TCS came to the radiology clinic at King Saud University for cone beam computed tomography examination (CBCT). Both father and older brother showed the same features of TCS as well. CBCT examination of the maxillofacial region was performed on the patient using a large field of view using a Carestream® CS 9300 CBCT machine. Sectional images along multiple planes were obtained, including cross-sectional images of the maxillofacial area.

Extraoral examination showed a small face with hypoplasia of the mandibular and zygomatic bones (Figure 1). The hypoplastic mandible and zygoma caused protrusion of the upper teeth, and the nose appeared very prominent. The patient's eyes were noticeable, and there was microphthalmia of the lower eyelid laterally as well as partial absence of the lower eyelid lashes. External ear malformation was also noted in the form of a bilaterally underdeveloped pinna (Figure 2). The patient was wearing a surgically implanted device just behind the right ear called (BAHA), which is a bone-anchored hearing aid that is designed to help people with deafness. It transfers sound through bone to the inner ear.

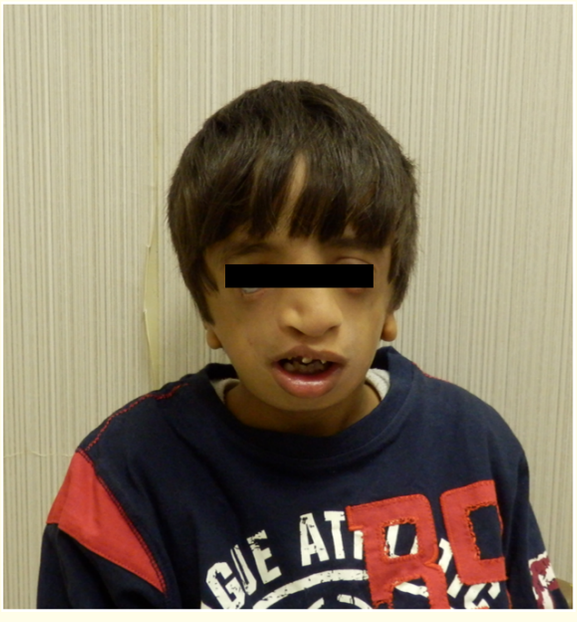

Figure 1: *Frontal facial aspects clinical of the patient showing the slants of the palpebral fissures, mandibular and zygomatic hypoplasia, incompetent lips and absence of lower eyelid cilia.*

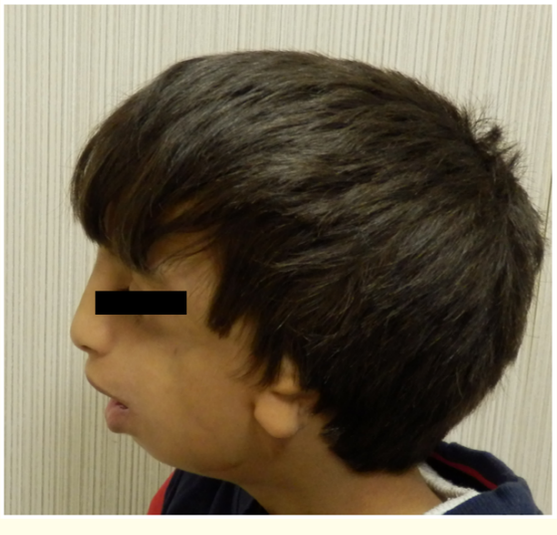

Figure 2: *Clinical features of deformities of the external ear often leading to conductive hearing loss. Micrognathia of the mandible is also notable.*

Radiographic examination revealed a hydrocephalic head with an underdeveloped maxilla and absence of the bilateral zygomatic arches (Figure 3). A small right maxillary sinus was also seen in relation to the contralateral side. Narrowing of the oropharyngeal airway space (Figure 4) was also observed. The left condylar head was absent, whereas the right condylar head was hypoplastic with bilateral deep antegonial notches (Figure 5). An enlarged inferior concha and deviated nasal septum were also noted (Figure 6). Overall, both radiographic and clinically discovered features were suggestive of TCS.

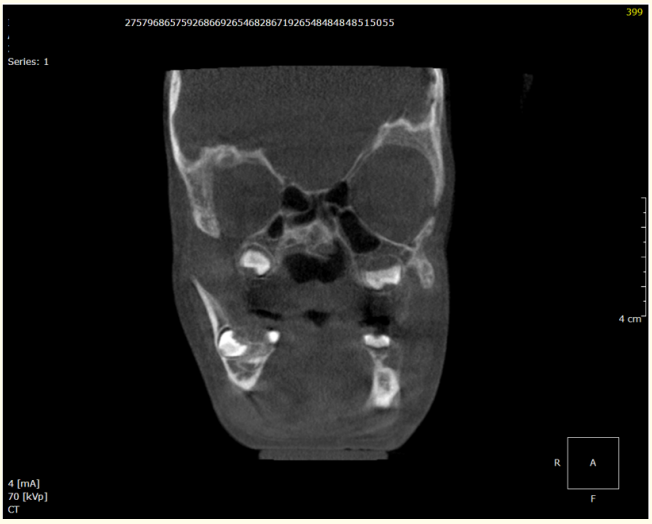

Figure 3: *Coronal view demonstrating small right maxillary sinus and absence of zygomatic arches bilaterally.*

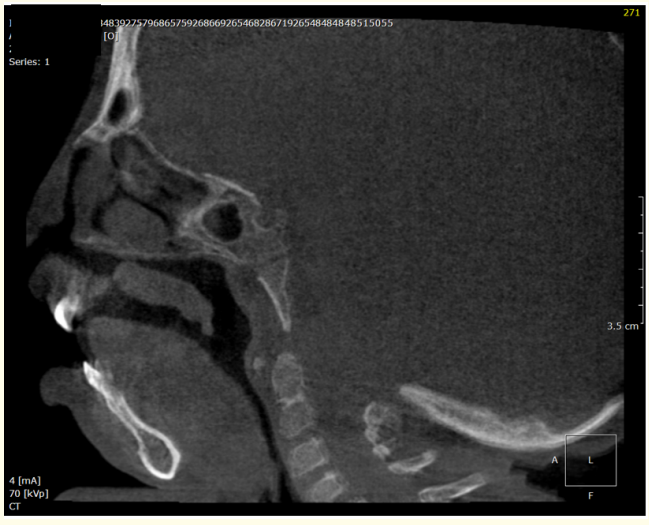

Figure 4: *Sagittal view showing narrow oropharyngeal airway space.*

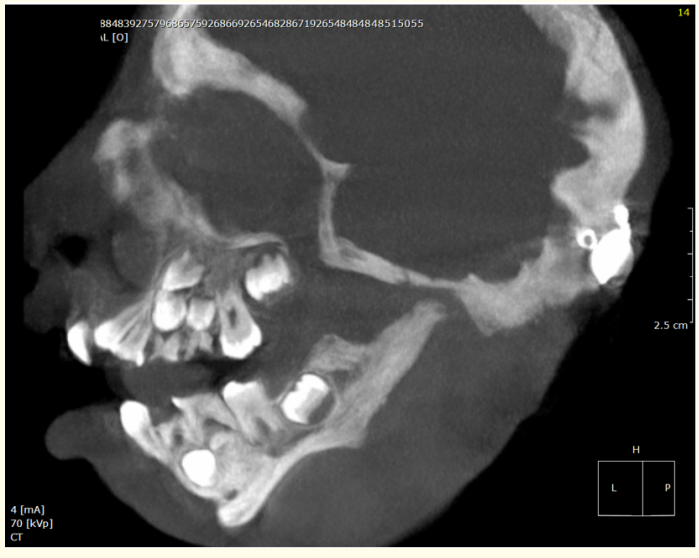

Figure 5: *Sagittal view showing absence of the condylar head, deep antegonial notch and BAHA.*

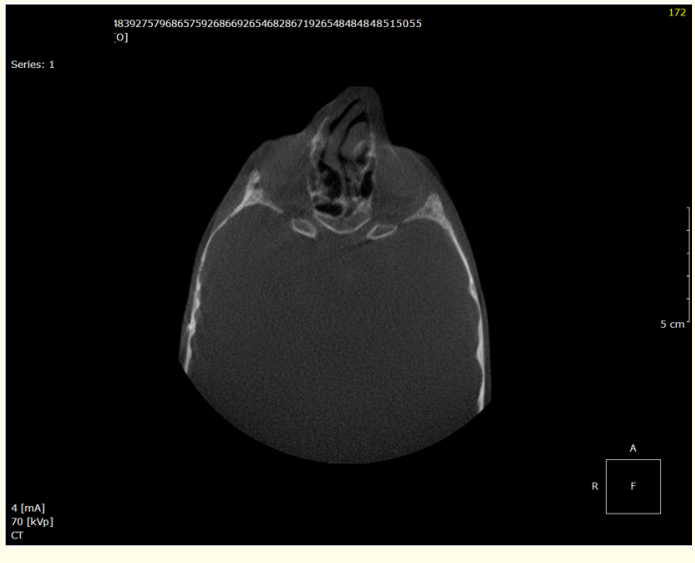

Figure 6: *Axial view showing deviated nasal septum.*

Discussion

TCS is a severe congenital developmental disorder characterized by multiple developmental anomalies in the craniofacial bones. Hypoplasia of the facial bones, especially the zygomatic complex and mandible, is a widespread feature of TCS [6]. It affects both sexes equally. Sixty percent of the discovered cases appear to have no family history, while the remaining forty percent are found to have a positive family history [7]. In our case, the patient had a family history of the syndrome.

TCS is characterized by bilateral external ear abnormalities, hypoplasia of the zygomatic bone, micrognathia and down slanting of palpebral fissures. In early life, feeding may be a significant challenge due to hypoplasia of the zygomatic bone and mandible. Malformation of the ossicles and hypoplasia of the middle ear cavities cause conductive hearing loss in approximately 40% - 50% of affected individuals. While inner ear structures found to be normal. Cleft palate and unilateral or bilateral choanal stenosis or atresia are other less common abnormalities [8]. Studies have demonstrated mild alterations in skeletal morphology and growth, with remarkable disturbances in maxillary and mandibular positioning and the tendency to preserve the syndromic facial aspect with time [9]. Dental findings include impacted supernumerary teeth in the maxillary anterior region, hypoplasia and position alterations of the maxillary central incisors [10]. Features observed less frequently in individuals with TCS include nasal deformity, macrostomia, high palatal vault and anterior open bite with class II malocclusion [8].

Computed tomography (CT) has been used to determine and document abnormal anatomic structures in syndromic and non-syndromic craniofacial anomalies. This helps to reach a correct treatment plan through good diagnosis. Furthermore, CT data also help in the assessment of the results obtained after surgical intervention for these complex disorders [10,11].

Conclusion

There is no cure for TCS. Treatment is aimed toward the specific needs of each affected individual. Many children require a multidisciplinary team involving maxillofacial surgeons, plastic surgeons, pediatric otolaryngologists, audiologists, geneticists, psychologists and other healthcare professionals. It is highly recommended to have genetic counseling for affected individuals and their families [12]. Consequently, more research should be directed toward the preventive aspects of this syndrome.

Conflict of Interest

There is no financial interest or any conflict of interest exists.

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