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A Unique Case of Unilateral Treacher-Collins Syndrome with Middle Ear Aplasia: A Case Report

Case Report

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Abstract

Treacher-Collins syndrome or Mandibulofacial dysostosis (MFD) is a rare congenital disorder with autosomal dominant inheritance, associated with aberrations of craniofacial development. The most common clinical features include malar bone and mandibular hypoplasia, antimongoloid slanting of palpebral fissures, and other ear abnormalities. Computed tomography is the imaging modality of choice for the morphological analysis of the craniofacial bones in individuals with complex facial deformities and to assist in the planning of surgical intervention. We present a case of unilateral mandibulofacial dysostosis with absent middle ear ossicles.

Keywords: Treacher-Collins; Middle Ear Ossicles; Computed Tomography; Zygomaticbone

Introduction

Treacher-Collins syndrome or Mandibulofacial dysostosis (MFD) or Franceschetti-Zwahlen-Klein syndrome is a rare congenital disorder of craniofacial development with autosomal dominant inheritance. Approximately 60%of cases of Treacher-Collins syndrome show a genetic abnormality in the form of a de novo mutation involving the treacle gene (TCOF1) on chromosome 5 with resultant interference in the development of first and second branchial arches. Clinical features most encountered are hypoplastic malar bone and lower jaw, auricular abnormalities, and antimongoloid slanting of palpebral fissures. [1] Computed tomography aids in diagnosing anatomical abnormalities, developing surgical treatment strategy as well as post-operative monitoring. [2] We report an interesting case of Treacher-Collins syndrome, discussing its clinical and computed tomography imaging features.

Case Report

A 10-month-old male, born out of a 2nd degree consanguineous marriage and uneventful antenatal and immediate post-natal history was brought to medical attention with complaints of deformed right pinna and regurgitation during feeding. On detailed clinical examination, the child had antimongoloid slant of eyes, narrow face with right mandibular hypoplasia, deviation of angle of mouth to the right (Figure 1-A), high arched palate, bifid uvula and retrognathism. External ear malformation was noted in the form of grade III microtia (Figure 1-B) on the right andabsence of an external opening of the right ear.Above features were suggestive of multiple craniofacial abnormalities.

High-resolution computed tomography (HRCT)of Temporal boneon 160 slice Multidetector CT scanner (MDCT) with thin sections revealed,type 3 microtia and an aplastic external auditory canal on right (Figure 2-A) and (Figure 2-C). Absent right middle ear ossicles with short and wide lateral semi-circularcanal (Figure 2-B) and (Figure 2-C). The facial canal at the level of the labyrinthine segment of facial nerve appeared narrowed as compared to left. Rest

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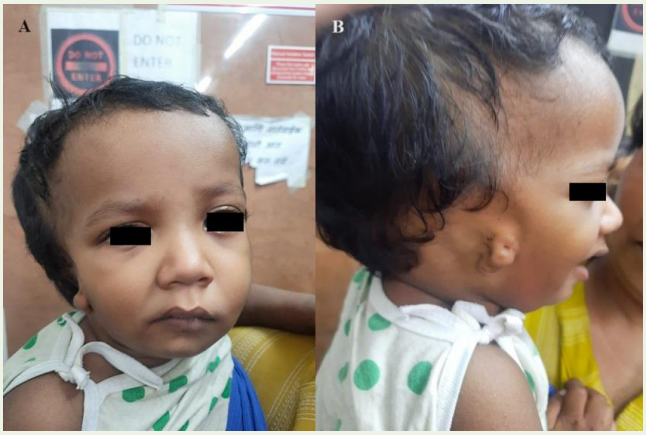

Figure 1: Frontal (A) and lateral (B) profile photographs of the patient demonstrate antimongoloid slant of the right eye, deviation of angle of mouth to the right, retrognathism, and microtia.

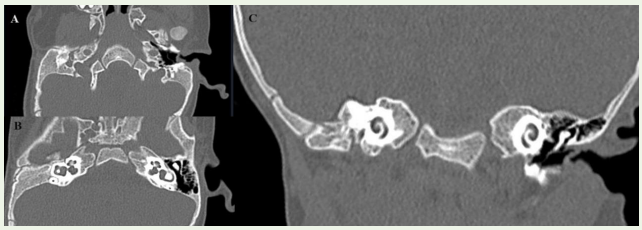

Figure 2: Axial non-contrast CT Temporal bone (bone window) with coronal MPR reconstruction reveals an absent right external auditory canal and non-pneumatized right mastoid (images A and C), absent right middle ear cavity, and ossicles (images B and C). There is a short and wide right lateral semicircular canal (B).

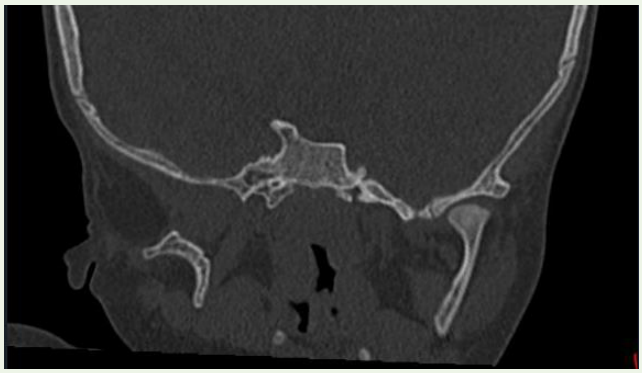

Figure 3: Sagittal MPR reconstructed section CT temporal bone shows absent or dysplastic right mandibular condyle.

of the facial nerve canal was not well appreciated. There was non-pneumatization of right mastoid air cells (Figure 2-A) and (Figure 2-C). Absent right zygomatic bone and condylar process of right mandible (Figure 4-A).

Informed consent was obtained from the father of the patient involved in the study and was informed about the potential publication in scientific journal.

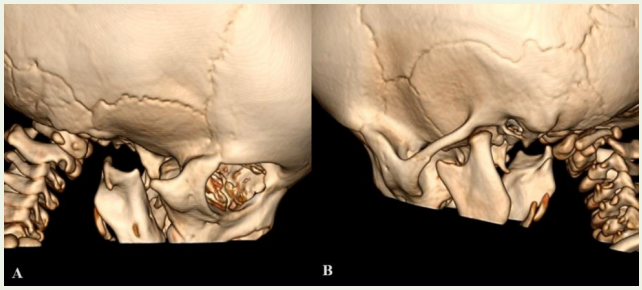

Figure 4: 3D CT Volume rendering technique (VRT) images, right lateral (A) and left lateral (B) view show absent or dysplastic right mandibular condyle and zygomatic bone (image A) and normal left mandibular condyle and zygomatic bone (image B).

Discussion

Treacher-Collin syndrome is a disorder of craniofacial development with an estimated incidence of 1 in 50,000 live births with positive family history in about 40% cases and new mutations in 60% cases.[3] A British ophthalmologist Treacher Collins first described the features of this anomaly in 1900, however the first case was reported nearly 54 years prior in 1846 by Thompson.The condition was named mandibulofacial dysostosis in 1944 by Franceschetti, who wrote a significant revision of the anomaly [1].

TCOF1 gene, which codes for serine and alanine-rich nucleolar phosphoprotein necessary for craniofacial development is identified as the gene responsible for this condition [4].

The clinical features originallydescribed by Franceschetti and Klein[5] include antimongoloid palpebral fissure, facial bone hypoplasia (predominantly malar bone and mandible), external ear deformity, occasional middle and inner ear abnormalities, dental abnormalities, high arches palate, and other skeletal deformities. Our caseshowed right sided microtia with absent external ear opening, antimongoloid slant of eyes, hypoplasia of the right mandible with a narrow face, right side deviated angle of mouth, high arched palate retrognathism, and bifid uvula. They also described five clinical forms of this syndrome: complete form, incomplete form, abortive form, unilateral form, and atypical form. Our patient had the unilateral form with involvement only on the right side.Tebe*et al.* defined hypoplasia of zygomatic arch and downward slanting palpebral fissures as minimum diagnostic criteria, both of which were seen in our case [3].

Absent, malformed, or malposed external ears and variable degrees of hypoplasia of the external auditory canals and middle ear ossicles were CT findings described by Posniak et al.[6] According to Stovin et al, the absence of a zygomatic arch was the chief radiologic finding. [7] On the basis of CT volumetric studies, Terner et al demonstrated that the mandibular condyle is the most common hypoplastic structure as compared to the rest of the mandible.[8] Loyd*et al in* 1979 described underdeveloped and under-pneumatized mastoid as the most obvious and constant feature in this syndrome. [9] MDCT of our patient revealed external ear malformation in the form of right-sided microtia and aplastic external auditory canal. All three right middle ear ossicles were absent. The left semi-circular

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canal was short and wide. Condylar process of the right mandibular and right zygomatic bone was absent. Also, the right mastoid air cells were non-pneumatised in our case. In our case, however, unilateral affection was noted, only on the right side.

Facial features similar to this condition are also seen in Nager's syndrome, Miller's syndrome, and Goldenhar syndrome hence must be included as a differential diagnosis of TCS. However, preaxial limb abnormalities and more severe mandibular hypoplasia favour Nager's syndrome. Miller's syndrome can be differentiated on the basis of ectropion and postaxial limb defectswhile features like anomalies of vertebrae, epibulbardermoids, and asymmetrical facial anomalies are characteristic of Goldenhar syndrome. [10,11]

For the management of this condition, a multidisciplinary approach is necessary which involves orthodontists, craniofacial surgeons, otolaryngologists, ophthalmologists, and speech therapists [12].

Mutation of the gene can be identified by genetic testing, however is expensive and hence is employed only if clinical findings are equivocal [13].

To reduce the incidence of this syndrome, prenatal diagnosis and genetic counselling of parents is recommended [14].

This case is unique, as it has unilateralinvolvement, whereas majority of the cases published in the literatureshowed bilateralinvolvement. Additionally, another uncommon finding in our case was the absence of all three middle ear ossicles.

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