CASE REPORT

Treacher Collins Syndrome: A Case Report and Review of Literature

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ABSTRACT

Treacher Collins syndrome or mandibulofacial dysostosis is a rare autosomal dominant disorder of craniofacial development. A case report of a 12-year-old girl having Treacher Collins Syndrome with depressed zygomatic arches, sunken cheekbones, on commissure area there scar was present, anterior open bite, posterior crowding was present and multiple impacted teeth in maxillary arch total teeth was 27 in maxilla. Different treatment options and diagnostic feature and mock surgery and CBCT was done to correct the disorder and psychological need to give a child a quality of the life.

KEYWORDS: Treacher Collins Syndrome, Mandibular Dysostosis, Crowding

INTRODUCTION

Treacher Collins syndrome (TCS) also called treacher Collins franceschetti syndrome or mandibulofacial dysostosis (MFD), is an autosomal dominant disorder of craniofacial development that occurs with an incidence range from 1:40,000 to 1:70,000 live births.[1] first dercribed by Treacher Collins (1900) and Franceschetti and Klein (1949). Treacher Collins Syndrome is caused by mutation of the TCOF1 and POLR1D genes. In the case of TCOF1 or POLR1D, is autosomal dominant, while of POLR1C is autosomal recessive. Clinical features of treacher Collin syndrome include midface hypoplasia, micrognathia, microtia, conductive hearing loss, cleft palate, widely spaced teeth, malocclusion, anterior open bite, and enamel hypoplasia. A diagnosis is made based upon a thorough clinical evaluation, a detailed patient history, and identification of characteristic findings. Specialized imaging techniques such as x-rays or computed tomography may be performed to assess the extent of certain craniofacial abnormalities.[2,3]

CASE REPORT

A 12-year-old girl reported to the Department of oral and maxillofacial surgery with a chief complaint of multiple impacted Teeth in the upper arch and facial esthetic. On clinical examination depressed zygomatic arches, sunken cheekbones, commissure area there scar was present, left lower eyelid downward, anterior open bite posterior crowding was present and multiple impacted teeth in maxillary arch total of 23 teeth. Clinically nasal septum was deviated towards left side (Figure 1).

Adequate mouth opening and there is deviation to right side while closing and opening the jaw. On intraoral examination anterior open bite with posterior crowding and pre mature contact of maxillary molars, high arched palate with a submucosal cleft and narrow arched maxilla. There are total 23 teeth present in maxillary arch with 4 impacted on the right side and 3 impacted on left side and dental caries was present in relation to 36,25 and 16 (Figure 1). Patient has a habit of mouth breathing and tongue thrusting.

Patient parents gave history of commisuroplasty and extraction of maxillary molar. Patient also gave history of delayed speech and nasal tawag were elicited from a detailed case history.

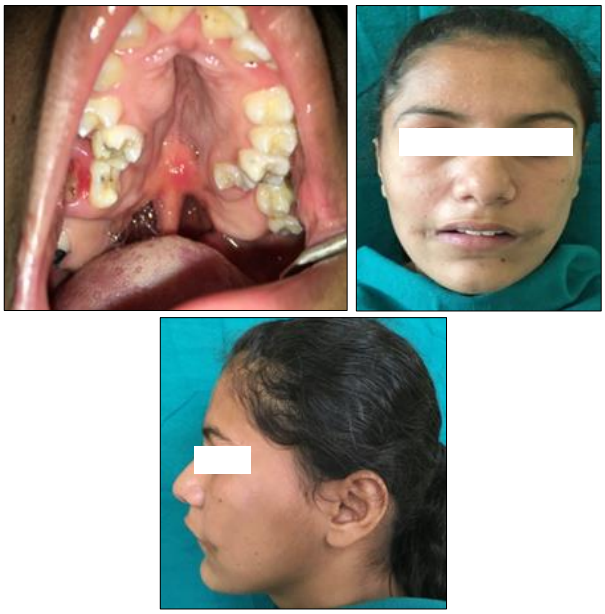

Figure 1:Clinical features of Treacher Collins syndrome patient

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Orthopantomogram of patient showed multiple impacted teeth in maxilla with total 25 teeth wa present with impacted mandibular third molar, hypoplastic zygomatic arches (Figure2).

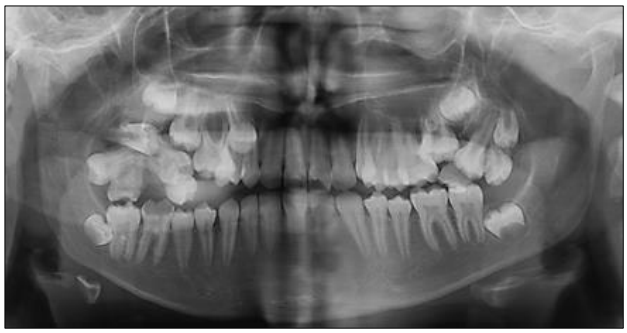

Figure 2: Orthopantomogram of patient

Length of the ramus is short and posterior crowding present in maxilla and crossbite with narrow maxilla. There was difficulty in swallowing and hearing and impaired vision and slurred speech and nasal tawang. CBCT reveals multiple impacted suparnumary and wisdom teeth present in maxilla and impacted teeth in mandibular arch. CBCT also reveals narrow arch with crowding and deficit hard palate (Figure 3).

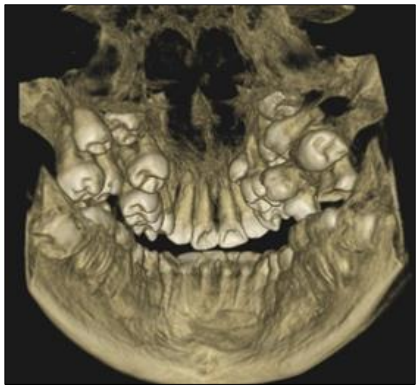

Figure 3: CBCT of patient

On the basis of clinical findings and radiographic findings the diagnosis of Treacher Collin Syndrome was made. The cephalometric analysis was done and showed reduced anterior cranial base length and decreased mandibular height. Impression was taken using alginate making a posterior stop and bulb to prevent alginate to fall into the pharynx and cast was made for diagnostic purpose and mounted on the hanau articulator for stimulation and mock surgery was performed to evaluate maxilla after removal of all impacted supranumary and wisdom tooth. Extraction of 5 supranumary teeth irt 17 and 3 supranumary teeth in maxilla, Restoration of 25,36 and maxillary arch expansion by quad helix and then followed by comprehensive orthodontic therapy was planned (Figure 4).

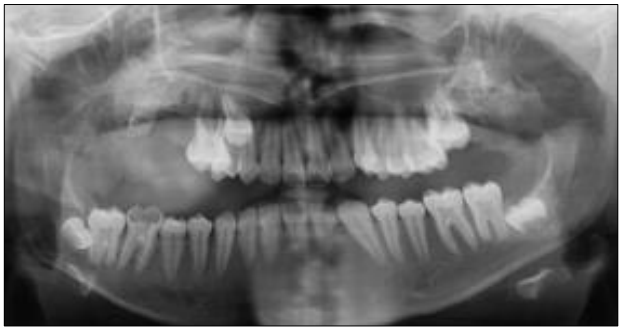

Figure 4: Orthopantomogram of patient

DISCUSSION

Treacher Collins syndrome is a autosomal dominant genetic disorder. It is caused by mutation of the TCOF1 gene encodes a nuclear phosphoprotein treacle, which exhibit linkage to human chromosome 5q32 locus. Recently Dauwerse *et al*. detected mutations in genes encoding subunits of RNA polymerases I and III in Treacher Collins syndrome patients. There is no family disposition in the patient with treacher Collin syndrome more than 60% patients. Our case doesn't show the positive family history suggesting familial mutation transfer in TCOF1 gene is absent. The side effect of autosomal dominant disease mutant genes observed in a large number of craniofacial syndromes craniosynostosis, maxillary hipoplasia, syndactyly-symphalangism, acne, brachymelia, velocardio-facial syndrome and Robinow syndrome.

In these syndromes there are majority of craniofacial multiple anomaly syndromes, there are both craniofacial anomalies and extracranial anomalies derived from the first and second branchial arches.Diagnostic features of Treacher Collin Syndrome include abnormalities in increased commisure, eyes, ears, impacted tooth, and depressed malar process. Based on these clinical features five clinical forms of Treacher Collin syndrome identified by Franceshetti and Klein.[7]

Clinical type of treacher Collin syndrome are 1) Complete form have all the features 2) Incomplete form present with less severe ear, eye, zygoma, and maxillary abnormalities 3) Abortive form in this only the lower lid pseudo coloboma and zygoma hypoplasia was present 4) Unilateral form unilateral anomalies on face and 5) Atypical form in which combined with other abnormalities not usually part of this syndrome. In our study, the patient presented the complete form of treacher collin syndrome. Kasat and Baldawa describe

Treacher Collin Syndrome as obligatory features of Treacher Collin syndrome given by Axelsson *et al*. in 1963 which include commissure area, antimongoloid palpebral fissures and anomaly of the lower eyelid, hypoplasia of zygomatic malar bone and hypoplasia of mandible or maxilla.[8]

The pathogenetic mechanisms involved in syndrome have been controversial and no definitive causal agent could be found so far. Treacher Collin Syndrome represents a defect of blastogenesis that could be attributed to interferences in cephalic neural crest cell histopathologically differentiation. Differential diagnoses are nager and miller syndrome and hemifacial microsomia and Goldenhar syndrome. Miller syndromes include ectropion or out turning of the lower lids and similar features of treacher Collin syndrome.[1,7] Hemifacial microsomia disorder affects the development of the ear and mandible. Goldenhar syndrome shows vertebral abnormalities, epibulbar dermoids and facial deformities.[2,9,10] There is an estimated 35% incidence of cleft palate with or without cleft palate and an additional 30% to 40% incidence of congenital velopharyngeal incompetence in TCS patients. Patient does not have hypoplastic thumb, fusion of radius and ulna, ectropion of lower lids, cleft lip, vertebral anomalies so a diagnosis of TCS was given.

Patient with TCS may have problem in deglutition due to inability of the pharynx to handle large pieces of food. Patients prefer soft, easy to chew foods, might connect this preference to the anterior skeletal open-bite characteristic of this syndrome. Early diagnosis of Treacher Collin Syndrome allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients to live a better life. Genetic counseling is highly recommended for affected individuals and their families.

CONCLUSION

Treacher Collin syndrome is an autosomal disorder with abnormalities structures derived from 1st and 2nd branchial arch. Every patient of Treacher Collin syndrome is different. Many abnormalities of these disease improved by surgical interventions. A proper planning and discussing making can produce excellent results for patient. When confirmed with Treacher Collin syndrome by clinical radio graphical and history by patient and parents it is important to pay attention to psychological need.

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