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RESEARCH ARTICLE

TREACHER COLLINS SYNDROME: A CLINICAL CASE REPORT

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ABSTRACT

Treacher Collins syndrome or mandibulofacialdysostosis is the condition where the cheek-bones and jaw-bones such as maxilla and mandible are under-developed. Treacher Collins is the diagnosis, which can be given to the children who have notching or stretched lower eyelids, it can be associated with partially absent cheekbones and small mandible. Here we report a case of treacher collins syndrome with a characteristic clinical presentation

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INTRODUCTION

Treachercollins syndrome (TCS) also known as mandibulofacialdyostosis syndrome or syndrome of Franceschetti is a hereditary disorder, which is characterized by craniofacial anomalies and numerous clinical manifestations (Bezerra *et al*., 2005). It has an incidence of 1 in 40000-70000 cases per live born children. No sex and racial predilection (Hungria, 1991). The defective gene TCOF1 is located in the distal portion of long arm of chromosome5 (5q31.3-q33.3) (Arn *et al*., 1993). Dr. Teacher Collins, a British opthamolgist was the first person to report a case of two children who had very underdeveloped cheekbones and notches in the lower eyelids in 1900. The disease is thought to occur due to the interference in the development of the 1st and 2nd brachial arches (Granstrom and Jacobsson, 1999; HereilioMartelli-Junior *et al*., 2009) and approximately 60% of the cases arises probably as a de novo mutations. Only the remaining 40% cases have a previous family history (Berry, 1889).The aim of this case report is to discuss clinical presentation of treachercollins syndrome and its modes of management.

Case report

An 18 year old female patient reported to our dental op with the chief compliant of pain in her right lower back tooth region for past 1 month.

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Anamnesis revealed that patient had undergone surgical correction of cleft palate when she was 3 years of age. Her facial profile(Fig 1) revealed asymmetry on left side of the face, increased inter-canthal distance, frontal bossing, deficient/hypoplastic malar bones, prognathic anterior maxilla, short upper lip, incompetent lips, absence of mental groove and retrognathic mandible(Fig 2).

Her ENT examination confirmed deviation of nasal septum towards left side anddeformity of the left ear with absence of left auditory canal, resulting in loss of hearing ability in the Left ear. Intra oral examination revealed (Fig 3) narrow and high arch palate with evidence of scar. All the permanent teeth were fully eruptedwith multiple dental caries. The loweranterior teeth were crowded andgeneralized enamel hypoplasia was evident.

The patient also presented with a deformed uvula with the bifid morphology (under developed in the left side). Radiographic survey with OPG and Lateral Cephalogram were done, of which the OPG revealed (Fig. 4) prominent antigonial notch on the left side, short ramus, hypoplasia of the left side of mandible. Lateral cephalogram revealed (Fig 5) hypoplasia of zygomatic bone andproclined upper anterior teeth and retrognathic mandible. Co-relating the clinical and radiographic features a provisional diagnosis of Treacher Collins syndrome was made.

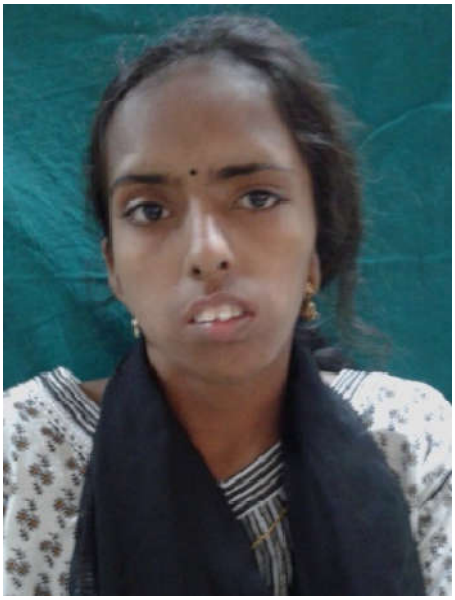

Figure 1. Profile Picture

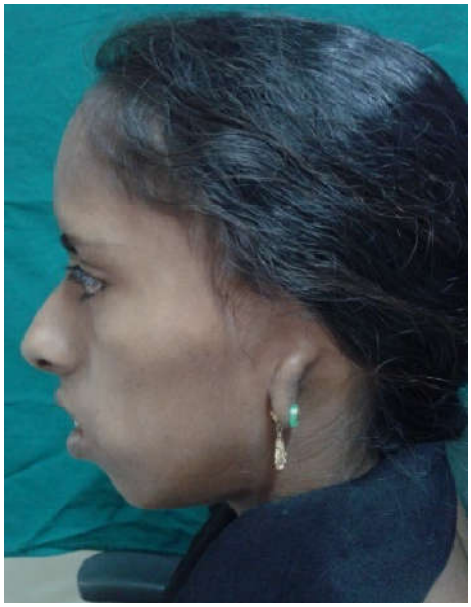

Figure 2. Side View

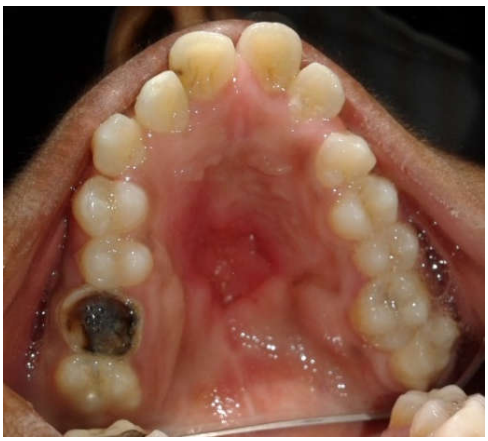

Figure 3. Intra oral picture

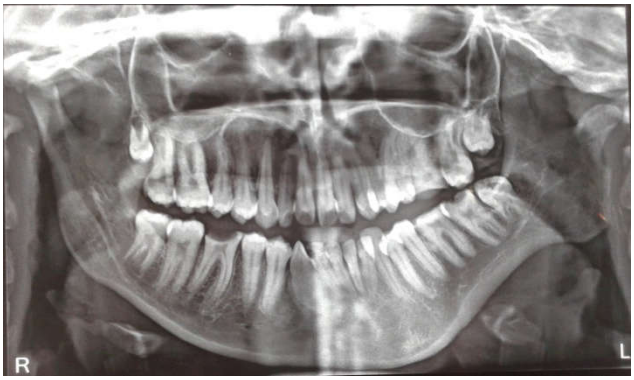

Figure 4. OPG

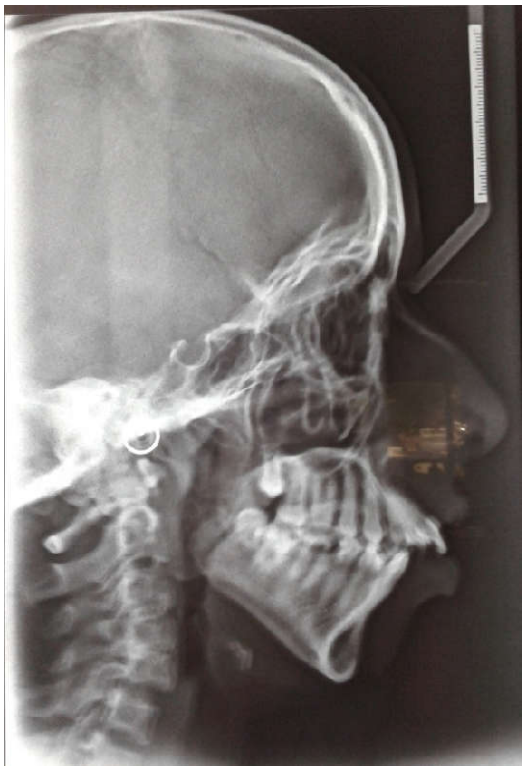

Figure 5. Lateral ceph

DISCUSSION

As mentioned earlier Treacher Collins Syndrome (TCS) is an autosomal dominant hereditary disorder, hence genetic factor plays a major role in its etiology. The mutation is specified to the TC0F1, POLR1C, and POLR1D.(Nord Guides for Physicians#12: The Physicians Guide to treachercollins syndrome)Interestingly, it is also found that TCOF1 and POLR1D has autosomal dominant inheritance, whereas the POLR1C has autosomal recessive inheritance. The TCOF1 gene is located in distal portion of long arm of chromosome5 (5q31.3-q33.3) (Wise *et al*., 1997; Dixon *et al*., 1991), genes POLR1C (6q21.2) and POLR1D (13q12.2) (Nord Guides for Physicians12: What causes Treacher collinssyndrome; The Physicians Guide to treacher collins syndrome) are located in chromosome 6 and chromosome 12 respectively. Other factors such as vitamin deficiency, malnutrient pregnant mother, coalescense of amnion strings, attempted abortions, radiation, pressure, uterine hemorrhage, variation in temperature,

diabetes, certain drugs, diptheria, toxoplasmosis, measles, rubella, influenza, maternal age, tumors, nervous shock, marital difficulties, socioeconomic conditions etc. are considered to have an etiological influence on TCS.

Treachercollins presents itself with various clinical presentations and varies in affected individuals, even if among family members (Rovin *et al*., 1954). The syndrome commonly affects the bones of the face, ears and orbit, and usually have a bilateral presentation. Respiratory complications may sometimes be fatal and the clinical presentation may be complicated due to the fact that respiratory ventilation is difficult to achieve due to defective facial bone development (Negamine and Kurahashi, 2007; Jayasekera, 2007). TCS presents either with ears that appear normal, or the external ears may be abnormally small or completely absent. Atresia or stenosis of the external auditory canals, conductive hearing loss (ranging from mild to severe) usually due to malformations of structures within the middle ear (Herberts, 1962). Our case also showed deformity of the left ear with absence of left auditory canal with associated conductive hearing loss too. Eye abnormalities can give a saddened facial appearance to the affected individuals. Lower eyelid abnormalities increases the risk of eye infection. Specific ocular eye defect associated includes coloboma, drooping eyelids, downward slanting palpebral fissures, partial orcomplete absence of the lower eyelashes, dacrostenosis ornarrowed tear ducts, also vision loss (Roy, 1978; McEnery & Brennemann, 1937; Poswillo, 1976). Hypoplasia of the malar bones and mandible are classic facial features of TCS which complied in our case too. The retrognathic mandible gave a relatively prognathic anterior maxilla which indirectly led to incompetent lips. Approximately 55 to 60% of individuals with TCS may develop dental abnormalities including tooth agenesis, widely-spaced teeth, malocclusion, enamel opacities, ectopic eruption of maxillary first molars, macrostomia. Other findings, which less frequently occur are: High arched palate, nasal deformity, cleft palate (McEnery & Brennemann, 1937; Poswillo, 1976). Our case had enamel hypoplasia, high archedpalate with cleft and deformed bifid uvula. In the differential diagnosis, one should consider the acrofacialdysostoses. Acrofacial dysostoses such as Nager syndrome and Miller syndrome have appearances that resembles that of Treacher Collins syndrome, but additional limb abnormalities occur in those patients. Hemifacial microsomia which primarily affects development of the ear, mouth, and mandible can be considered a differential Diagnosis, however this anomaly may occur bilaterally. Another disease which belongs to this spectrum is Goldenhar syndrome, which includes vertebral abnormalities, epibulbardermoids and facial deformities. The classic ocular presentation in Goldenhar syndrome, ruled out this differential diagnosis hence confirming the initial diagnosis of Treacher Colins Syndrome.

Treatment

The variable clinical manifestation of treachercollins syndrome demands for precise management of the symptom or defect or deformity depending on the severity of the syndrome. Usually after birth, the child with treachercollins syndrome should be evaluated for airway and secure it at the earliest (Jayasekera, 2007). Failing to achieve airway will lead to death. In case of cleft deformity of the lip or palate the sequence of corrections starting from 3 months of age with appropriate follow ups are mandatory. Mandibular deficit correction and treatment for choanal atresia (a congenital disorder, which presents with narrowing or blockage of nasal airway by tissue) are done at 2-3years of age (Harrison, 1951; Andrade *et al*., 2005). Grafts like calvarial, ribs and iliac bone should be used. Autogenous graftare more preferred than synthetic graft. Even though the cranioorbito-zygomatic bone development is almost complete by 5-7years of age, reconstruction must be peformed only after 10 yrs of age (Harrison, 1951). Procedures like chin augmentation and mandible distraction are recommended to be performed before the completion of facial growth, keeping in mind that orthognathic surgery should be delayed until the age of 16 to 18 years. The correction of malocclusion should be linked hand in hand and other defects such as enamel hypoplasia which occurred in our patient can be managed with conservative methods in order to preserve the tooth structure. Tessier method is used to correct coloboma of lower eyelid; which includes z-plasty for cutaneous lengthening, and overlapping sutures of the preseptal orbicularis oculi muscle and canthopexy (Tessier and Tulasne, 1986).

Conclusion

From the above discussion it is very clear that Treachercollins syndrome is a rare manifestation and needs more focus on, improving the diagnostic ailments, as the critical period of pathogenic activity for deformities occurs around 7 weeks in utero, and to improve the current approach in treatment modalities from aesthetical correction to psychosocial development and also to bring in the multidisciplinary team of plastic surgeons, maxillofacial surgeons, otorhinolaryngologist, opthalmologist, speech therapist and psychologist in order to offer better quality of life to patients as well as their families.

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