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CASE REPORT

TREACHER COLLINS SYNDROME – A CASE REPORT

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ABSTRACT

Treacher Collins syndrome (TCS) is also known as Franceschetti-zwahlen- klein syndrome is a rare genetic disorder characterised primarily by craniofacial abnormalities such as deafness, hypoplasia of facial bones (malar, maxillary and mandibular bone), antimongoloid slant of palpebral fissure, coloboma of lower eyelid. It was first reported by Allen Thomson in 1846than in 1899 by George AndreasBerry, Edward Treacher Collins in 1900 and in 1949 by Franceschetti-zwahlen- klein. Here we present a case report that describes a clinical and radiographic characteristic of TCS in a 27 years old patient who had reported to our Oral Medicine & Radiology Dept.

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INTRODUCTION

The Mandibulofacial Dysostosis syndrome or Treacher Collins syndrome covers a group of defects of head and face. It is often inherited as autosomal dominant trait and is derived from the 1st and 2nd branchial arches between 11 – 14 weeks of gestation (Dalben *et al.,* 2006). The frequency is estimated to be 1 in every 50,000 new born babies. The gene for TCS was mapped to chromosome 5q32-5q33.1. The involved gene may include TCOF1, POLR1C or POLR1D. Molecular analysis revealed that TCOF1 insufficiency leads to decrease in neural crest cells, there by altering craniofacial development. Although TCS was first reported by Thomson in 1846 but was extensively described by Franceschetti-zwahlen- klein in 1949 who gave the term Mandibulofacial Dysostosis to describe the clinical features that includes hypoplasia of facial bones, malformation of ear, conductive deafness, cleft palate & nasal deformity (Dixon, 1995). The present case report illustrates the orofacial features of TCS.

CASE REPORT

A 27 year old male patient (Figure 1) reported to our Department of Oral Medicine & Radiology with a chief complaint of rough feeling in the right side of the tongue.

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On extra oral examination patient revealed a narrow face with hypoplastic mandible and hypoplasia of malar prominence along with antimongoloid slant of the palpebral fissures. The patient's eyes were remarkable and there was partial absence of lower eyelashes and coloboma of lower lateral eyelid (Figure 2). Unilateral presence of rudimentary pinna was evident and external ear canal was atresic along with conduction deafness (Figure 3). Nose appears very prominent. Hypoplastic maxilla was also evident. Intra oral examination revealed as anterior open bite, high arch palate, macrostomia, labially tilted tooth irt 13 (Figure 4), sharp lingual cusp irt 46 47, gingival recession irt 33 42 43 (Figure 5). Inflamed and enlarged foliate papillae were evident in the right lateral surface of the tongue (Figure 6). Radiographic examination of Orthopantomograph revealed prominence of antigonial notch, short ramus, hypoplasia of mandible, steep mandibular angle, mesioangular impacted tooth irt 38 and horizontal impacted tooth irt 48 (Figure 7). Based on patient's chief complaint he was first diagnosed with Foliate Papillitis and correlating all clinical and radiographic findings, a second diagnosis of Treacher Collins Syndrome was made. The patient was treated for his chief complaint and was prescribed chlorhexidine mouthwash(0.12%) to be used twice daily to promote good oral hygiene, was advised to avoid hot and spicy food, was prescribed topical lignocaine anaesthetic gel to be applied 3-4 times daily, Iron, folic acid, vitamin supplements were given and was referred to Dept. of Periodontics for Coronoplasty. He was also motivated for orthodontic treatment and genetic counselling was given to the patient.

Table 1.

Diagnostic criteria for Treacher Collins Syndrome		
		Present Case
Region	Characteristic Feature	
Eyes	Antimongoloid slant of palpebral fissures	+
	Colobomata and hypoplasia of lower eyelid and lateral canthi	+
	Hypertelorism	-
	Partial absence of eyelid cilia	+
Ears	Microtia	+
	Conductive hearing loss	+
	Hypoplasia of middle ear ossicles	+
Nose/	Nasal deformity	+
Mouth	Cleft palate with or without cleft lip	-
	High arched palate	+
	Class II or III malocclusion	+
Facial bone	Hypoplasia of the malar bones	+
formation	Hypoplastic lateral aspects of orbits	-
	Variable effects on the temporomandibular joints	-
	Anterior open bite	+
	A steep occlusal plane	+

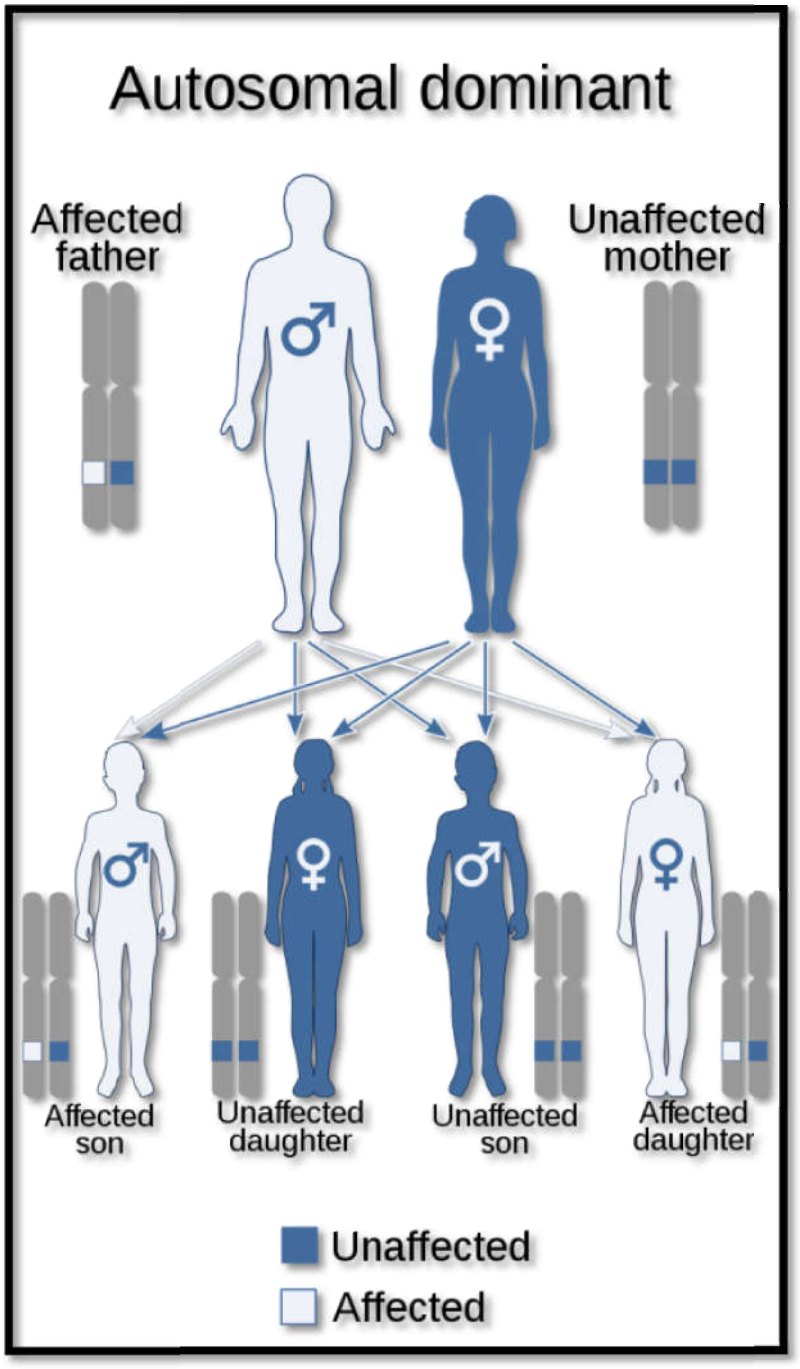

Chart 1.

Fig. 1. Patient Profile Picture

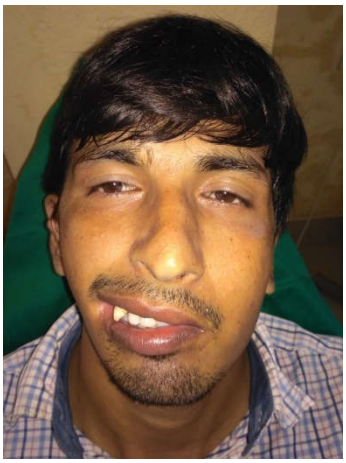

Fig. 2. Coloboma of lower eyelid

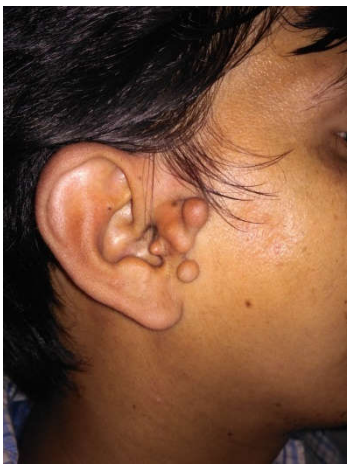

Fig. 3. Rudimentary Pinna

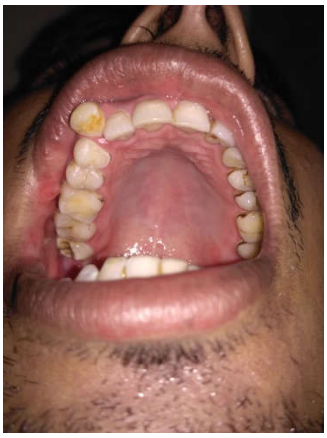

Fig. 4. Labially tilted tooth irt 13

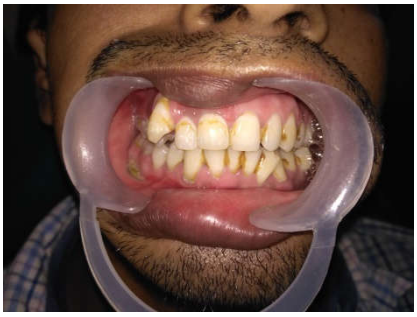

Fig. 5. Gingival Recession

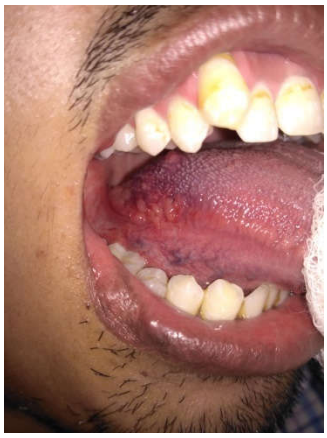

Fig. 6. Foliate Papillitis

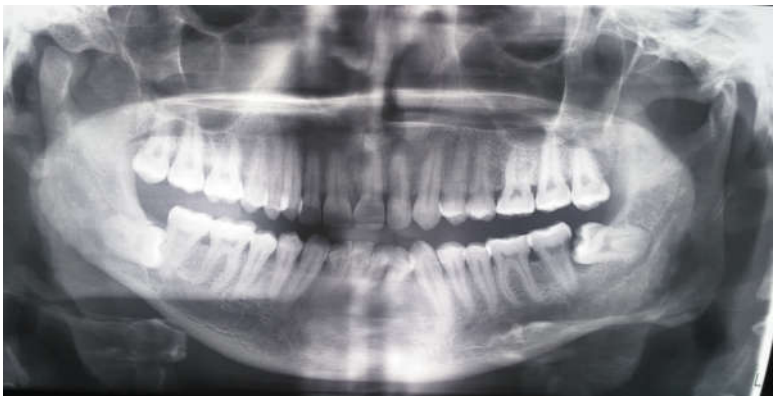

Fig. 7. Orthopantomograph

DISCUSSION

In 1846, Allen Thomson was the first scientist who refer to this syndrome. In 1900, Dr. Edward Treacher Collins, who was an surgeon and ophthalmologist, described two child patients with zygomatic hypoplasia in his case report, since then this condition has been termed as Treacher Collins Syndrome. TCS is transmitted by an autosomal dominant mode of inheritance. Mutation in TCOF1, POLR1C or POLR1D gene can cause TCS in which TCOF1 is the primary gene that is associated with TCS and being found in 90-95% of cases. TCOF1 is found on the 5th chromosome in the 5q32 region and mutation leads to haploin sufficiency of the treacle protein and loss of one copy of treacle affects the ability of cells to form facial bones and tissues (Valdez *et al.,* 2004).

A child may develop TCS in two conditions, first when both parents are normal and pass normal genes to the offspring but due to mutation the offspring develops TCS and the second one is that one of the male or female parents already suffer from this condition and pass their mutated gene to their offspring that results this condition (Chart 1). Although 40% of cases have present familial history and the remaining 60% appears to arise as a result of de novo mutation. In our case patient had no family history. Mann & Kilner assumed the etiology to be an inhibitory process occurring towards the 7th week of gestation and affecting facial bones and structures deriving from the 1st brachial arch. John Mckenzie suggested that the cause of this condition is malformation of stapedial artery during embryogenesis. Stapedial artery dysfunction gives rise to defects of the stapes and incus and the first arch vessels

supplying the maxilla. Failure of the inferior alveolar artery to develop an ancillary vascular supply gives rise to mandibular abnormalities (John Mckenzie and John Craige, 1955).

Franceschetti and Klein categorized into five clinical forms:

- The complete form (having all features)
- The incomplete form (presenting with less severe ear, eye, zygoma and mandibular abnormalities)
- The abortive form (only the lower lid pseudo-coloboma and zygoma hypoplasia are present)
- The unilateral form (anomalies limited to only one side of the face)
- The atypical form (combined with other abnormalities not usually part of this syndrome) (Hertle *et al.,* 1993).

In 1963 Axelsson include obligatory features that were antimongoloid palpebral fissures, anomaly of the lower eyelid, hypoplasia of malar bones and mandible. In our case patient presented with unilateral form of syndrome. Characteristics features of TCS include abnormalities in eyes, ears, nose, mouth and facial skeletons. Majority of these features were present in our case (Table 1). Nagar syndrome, Miller syndrome, Oculoauriculovertebral spectrum and Goldenhar syndrome should be included in the differential diagnosis of TCS. Nagar syndrome has similar facial features to TCS, especially in the region o eyes. Though, the mandible is more hypoplastic, preaxial limb abnormalities such as aplastic thumbs, fused radius and ulna are consistent features of Nagar syndrome unlike TCS (Chemke *et al.,* 1988). Miller syndrome also has some similarity in the facial features to TCS, in addition it has postaxial limb defects and ectropion of the lower lids and also cleft lip with or without cleft palate that is more common than in TCS (John Mckenzie and John Craige, 1955). Oculoauriculovertebral spectrum primarily affects development of ear, mouth and mandible (Kothari, 2012). Goldenhar syndrome shows vertebral abnormalities, epibulbardermoids and facial deformities (Shete *et al.,* 2011). There is no cure of TCS. Management of individuals affected by TCS requires a multidisciplinary approach involving craniofacial surgeons, orthodontics, ophthalmologists, otolaryngologists and speech pathologists. Depending upon clinical manifestations and severity, management may require tracheostomy at birth, multiple surgeries to correct eyelid coloboma and cleft palate followed by orbital reconstruction and maxillomandibular osteotomies, early recognition of deafness and its correction with hearing aids or surgery (Trainor *et al.,* 2009). Genetic counselling is highly recommended for the affected individuals and their families.

Conclusion

Each case of TCS is unique and requires to be assessed individually and in our case patient presented with unilateral form of TCS. Many features of this syndrome can be corrected and improved by surgeries and supportive treatments those results excellent outcomes for complete restoration of the form and function of the patient.

Patient Consent: Obtained

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