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RESEARCH ARTICLE

FAMILIAL TREACHER COLLINS SYNDROME – REPORT OF TWO RARE CASES WITH REVIEW OF LITERATURE

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ABSTRACT

Treacher Collins syndrome (TCS) or Franceschetti syndrome is an autosomal dominant disorder of craniofacial development affecting the structures derived from the first and second branchial arches with variable phenotypic expression. It presents with a characteristic facial appearance which is easily recognizable. We report a case with a review of a Familial Treacher Collin Syndrome affecting father and son showing all the characteristic features of the syndrome.

Key words:

Treacher Collins Syndrome, Franceschetti Syndrome, Mandibulofacial dysostosis, Cleft Palate, Deficient Zygomatic

INTRODUCTION

Treacher Collin syndrome also known as mandibulofacial dysostosis and Franeschetti –Zwahlen – Klein syndrome is a rare autosomal dominant disorder of the craniofacial structures, the incidence of which is estimated to be 1 in every 50,000 live births.[1]Although this disorder was first described by Thomson (1846), Tonybee (1847) and Berry (1889), it is eponymously named after Edward Treacher Collins, a London ophthalmologist who first described it in the medical literature in 1900.[2]

The first extensive review of the condition was detailed by Franceschetti and Klein in 1949, who used the term mandibulofacialdysostosis to describe the clinical features. It is thought to arise as a result of mutation in TCOF1 gene which is responsible for craniofacial development. 60% of the cases of TCS are thought to arise denovo and 40% are thought to be familial. TCS is characterized by hypoplasia of the zygomatic complex, downward slanting of the palpebral fissures, mandibular hypoplasia, cleft palate, anterior open bite, malformed external ear, coloboma of the lower eyelid, hearing loss etc.[1,3] We report a case of familial Treacher Collins syndrome with almost all the features of the syndrome present.

Case report

A 17 year old male patient reported to department, wanting a plate to be made for his upper jaw as he was not able to swallow food or drink anything. His parents give a history of him having cleft palate since birth for which he had undergone two corrective surgeries. The first surgery was done when he was 1 ½ year old and the second was done when he was 13 years old. Even though the surgeries have been done there is still a small hole in the palate which caused difficulty in swallowing and drinking. Whenever he tried to drink anything it would come out from his nose.

He gave a history of congenital difficulty in hearing along with cleft palate for which he wears a hearing aid which is attached to his head since his childhood. Extra oral examination of his face revealed a prominent nose, downward slanting of the palpebral fissures of both the eyes, coloboma of the lower eyelids, absence of eyelashes on the lower eyelid medial to the defect, malformed auricles, deficient midface suggesting absence of the zygomatic complex, prognathic maxilla and retrognathic mandible. (fig. 1a) Intraoral examination showed the presence of permanent dentition with missing 27. A well-defined ovoid defect was seen in the anterior hard palate in the midline in line with the first premolars.

The defect measured approximately 8mm x 5mm. (Fig 1b) the soft palate appeared to be malformed and the uvula was shifted on the left side. The defect showed fogging on the mirror on mirror test suggesting an oroantral communication. (Fig 1c) He had an anterior open bite with angles class III subdivision with crossbite on the left side. (Fig 1d) Looking at the classical

features a provisional diagnosis of Treacher Collins syndrome was made.

Figure 1 a) front and lateral profile b) cleft palate c) malformed soft palate with shifted uvula d) anterior openbite

To further be sure of the findings orthopantomograph (O.P.G), lateral cephalogram, P. A Watters were made. The O.P.G showed the presence of permanent dentition, short ramus and prominent antegonial notches. (Fig 2c) The lateral cephalogram showed anterior open bite, short ramus, absence of zyoma, prominentantegonial notches, retrognathic mandible and reduced pharyngeal space. (fig. 2a) P.A Watters showed the complete absence of the zygomatic bone and zygomatic arch and hypoplastic maxillary sinus.(fig. 2b) All the above findings pointed straight to confirm the diagnosis of Treacher Collin syndrome. The patient was then referred to the respective department for necessary treatment.

Family history revealed the congenital hearing defect of the father for which he had undergone a surgery few years back. A thorough examination of the father showed a prominent nose, downward slanting of the palpebral fissures of both the eyes, coloboma of the lower eyelids, absence of eyelashes on the lower eyelid medial to the defect, deficient midface suggesting absence of the zygomatic complex, prognathic maxilla and retrognathic mandible. (Fig 3) Same radiographs (fig 4) as made above showed mild features as compared to the son but suggestive of Treacher Collins Syndrome.

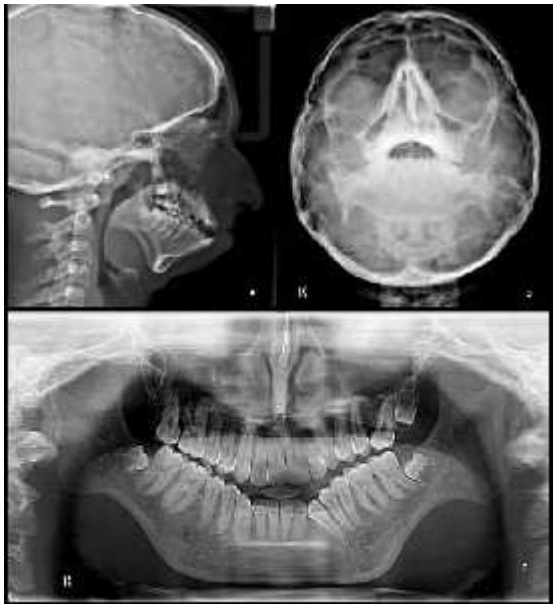

Figure 2 a) lateral cephalogram b) P.A. Watters c) Orthopantomograh

Figure 3 front and lateral profile of the father

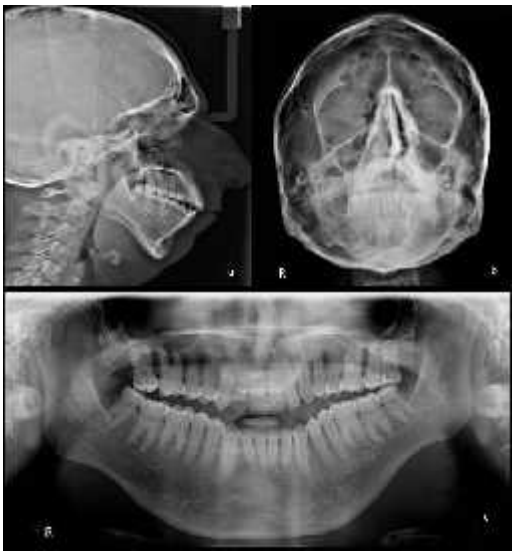

Figure 4 a) lateral cephalogram b) P.A. Watters c) Orthopantomograh of the father

DISCUSSION

From the structures affected and from studies in mice exposed to teratogeniccis or trans‑retinoic acid, it has been deducted that the disease results from interference in the development of the first and second branchial arches (Gorlin *et al*. 1990). [4]Franceschetti and Klein (1949) reviewed the literature and described the typical characteristics of the syndrome as follows: 1) Antimongoloid palpebral fissures with either a notch or coloboma of the outer third of the lower lid, and occasional absence or paucity of the lashes of the lower lid. 2) Hypoplasia of the facial bones, especially the malar bones and mandible. 3) Malformation of the external ear, and occasionally of the middle and inner ear, with low implantation of the auricle. 4) Macrostomia, high palate, malocclusion and abnormal position of the teeth. 5) Atypical hair growth in the form of tongue-shaped processes of the hair-line extending towards the cheeks in the pre-auricular region. 6) Association at times with other anomalies, such as obliteration of the naso-frontal angle, pits or clefts between the mouth and ear, and skeletal deformities.[5]After publication of this description, some of these features were regarded as being of lesser importance and some were emphasized in the diagnosis.

Thus, Axelsson *et al* (1963) named the following features as "obligatory": [5]

- Antimongoloid palpebral fissures
- Anomaly of the lower lid: coloboma of the outer third, or deficient lashes, or both
- Hypoplasia of the malar bones
- Hypoplasia of the mandible[5]

In our case, all the obligatory features as described above were present in the son.

Franceschetti and Kleinhasdescribed five clinical forms of TCS as: (i) the complete form (having all known features), (ii) the incomplete form (presenting variably with less severe ear, eye, zygoma, and mandibular abnormalities), (iii) the abortive form (only the lower lid seudocoloboma and zygoma hypoplasia are present), (iv) the unilateral form, (anomalies limited to one side of the face), and (iv) the atypical form (combined with other abnormalities not usually part of the typical syndrome).[6] In our case the son presented with the complete form and the father with the incomplete form. The differential diagnosis should include acrofacialdysostosis (Nager and Miller syndrome) and oculoauriculovertebral spectrum (hemifacialmicrosomia and Goldenhar syndrome). Nager syndrome has similar facial features to TCS, particularly in the region of the eyes i.e. downward slanting of the palpebral fissures with a deficiency of eyelashes. However, the mandible is usually more hypoplastic, the lower lid colobomas are rare, and preaxial limb abnormalities (hypoplastic, or aplastic thumbs, fused radius and ulna) are a consistent feature of Nager syndrome, which are not present in TCS[7].

Miller syndrome also has some similarity in the facial features to TCS; in addition it has postaxial limb defects (absence or incomplete development of the fifth digital ray of all four limbs) and ectropion or outturning of the lower lids. Also cleft lip, with or without cleft palate, is more common than in TCS and some patients may present with congenital heart defects.[8] Hemifacialmicrosomia primarily affects the development of the ear, mouth, and mandible but is usually unilateral while TCS is bilaterally symmetrical. Goldenhar syndrome shows vertebral abnormalities, epibulbardermoids and facial deformities.

There is no treatment for Treacher Collin Syndrome. Treatment aims at resolving the chief complaints and involves a multidisciplinary approach and the patients undergo a series of reconstructive and prosthetic surgeries.

CONCLUSION

Every case of TCS is unique and be assessed individually. Prenatal diagnosis of TCS in families with a history of TCOF1 using either fetoscopy or ultrasound imaging in the first trimester of pregnancy plays an important role. Research should be undertaken to find some preventive measures which when administered prenatally will prevent its occurrence. Early diagnosis of TCS allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients. If this can be done early, it is possible to take advantage of anticipated growth during normal skeletal maturation and to obtain better therapeutic results.

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