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Case Report

Maxillofacial Management of a Female Patient with Treacher Collins Syndrome – A Case Report from Egypt

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Abstract

Treacher Collins Syndrome (TCS) is a rare congenital disorder which causes multiple craniofacial deformities. TCS exhibits rare autosomal dominant inheritance [1], the symptoms are variable from severe life threatening conditions (airway obstruction that may lead to prenatal death) [2] to unnoticeable symptoms that can't be diagnosed on clinical basis only [3,4]. This is a case report of a 14 years old female patient from Egypt with Treacher Collins Syndrome who was brought to Oral and Maxillofacial Surgery Department at Faculty of Dentistry Suez Canal University to Extract remaining badly decayed deciduous teeth.

Keywords: Treacher Collins Syndrome; Oral and Para-Oral Deformities

Introduction

Treacher Collins Syndrome (TCS) is a rare condition was described for the first time by Thompson in 1846 [5], but it was named after the British Ophthalmologist Edward Treacher Collins in the year 1900 [6] who firstly described the essential features of the syndrome.

Later on, Franceschetti and Klein described the syndrome in 1949, in which they used the term mandibulofacial dysostosis to describe the facial hypoplasia [7]. It occurrs in one in 50,000 births, with equal prevalence in males and females, in 60% of cases arises from spontaneous mutation while in 40% of cases it's familial inherited [8].

It is a result of malformation of the first and second branchial arch that affects the shape and size of ears, eyes and facial bones [9].

It arises as a result of mutation in the TCOF1 gene (on chromosome 5q31.3-32) which encodes a rich nuclear phosphoprotein known as Treacle responsible for the craniofacial formation and development [10]. In 50% of individuals who have this mutation can pass it to their children [11]. Before identification of TCOF1 gene, diagnosis of this syndrome was only achieved by clinical examination and also by linked polymorphic markers (Dixon et al, 1994).

This syndrome is characterized by hypoplasia of facial bones, particularly the mandible (78% of cases) and zygomatic complex (81% of cases) [12].

Case Report

A 14 years old female patient came to Oral and Maxillofacial surgery department suffering from severely decayed deciduous teeth. the extra oral examination of the patient showed the characteristic features of Treacher Collins syndrome. she had cleft lip and palate and gone through cleft lip repair surgery at the age of 4 months and cleft palate repair surgery at the age of 9 months, she had very constricted undeveloped maxilla and normal sized mandible (figure 4), malpositioned palatally erupted permanent maxillary teeth, badly decayed retained upper deciduous teeth, badly decayed upper right first molar (figure 5), crowded lower permanent teeth and retained lower deciduous canines (figure 6).

Radiographic examination (Orthopantomogram) showed multiple impacted permanent teeth (figure 7). The patient suffers from cerebral atrophy, loss of vision in one eye due to malformation defect of cornea (figure 1,2,3) and bowlegs.

Family medical history

The mother is hypertensive patient and used to take anti- hypertensive drugs during pregnancy, the mother also was exposed to chest x-ray in her 7th month of pregnancy, the mother had abortion at the second month of her first pregnancy. The younger brother is autistic patient.

The whole family lives in a poor neighborhood close to chemical factories that release vapors in this area.

Treatment was carried out under general anesthesia, all badly decayed deciduous teeth were extracted along with the badly decayed upper right first molar then the case was referred to Orthodontic Department for retraction of permanent impacted teeth and proper alignment of maxillary and mandibular teeth.

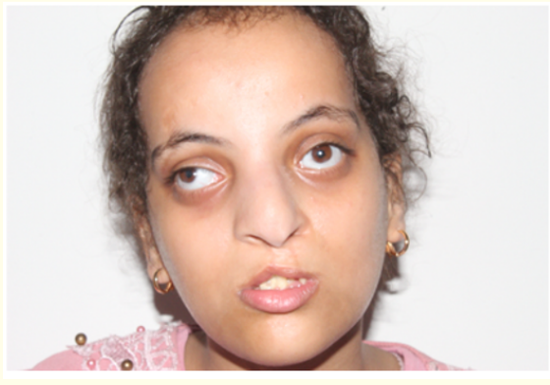

Figure 1

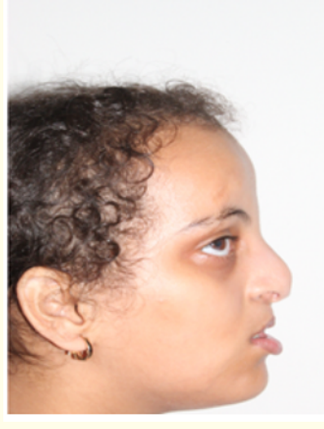

Figure 2

Figure 3

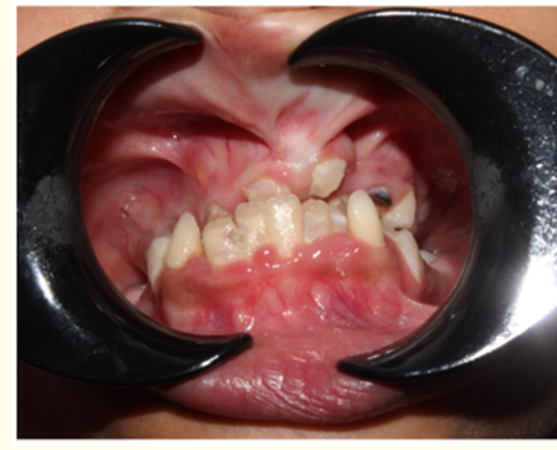

Figure 4

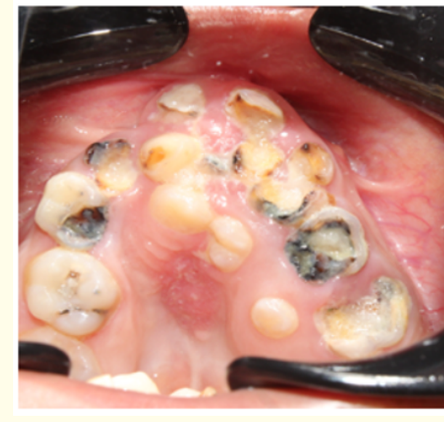

Figure 5

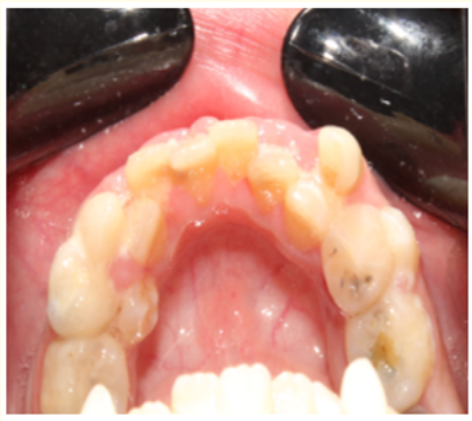

Figure 6

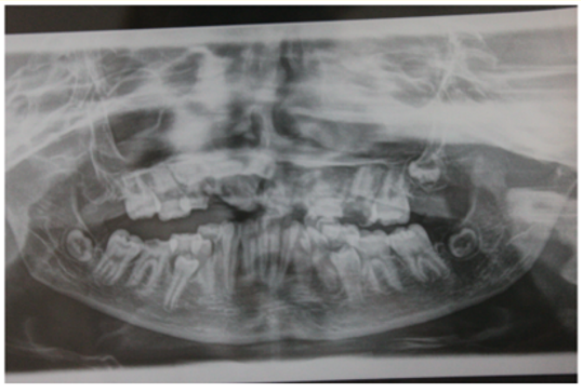

Figure 7

Discussion

Treacher Collins syndrome is rare autosomal dominant congenital condition that is characterized by oral and craniofacial deformities.

Franceschetti classified this syndrome according to the severity of its features into 5 categories: complete in which all features are present, incomplete with less noticeable features, abortive that shows hypoplastic zygomatic arch and coloboma, unilateral form that shows features only in one side and atypical form that shows abnormalities that are not included as features of the syndrome [13].

The clinical features of TCS in most of cases are bilateral and symmetrical, It affects oral and paraoral structures as well as other body organs, these features include:

Oral and Para-oral manifestations

- Hypoplasia of zygomatic complex including maxilla (81% of cases) and hypoplasia of mandible (78% of cases) [12], in the presented cases there is hypoplasia in the zygomatic complex and maxilla with normal mandible.
- Cleft palate (28% of cases) [14], the presented case had cleft lip and palate that were surgically corrected (cleft lip at the age of 4 month and cleft palate at the age of 9 months).
- Malocclusion and anterior open bite due to mandibular retrognathia [15] but in the presented case there is no anterior due to the normal size of mandible.
- Shortened soft palate, and some cases show bifid uvula.
- Enamel hypoplasia.

Other body organs may be affected as well

- Ears: Abnormalities of the external ears leading to hearing impairment.
- In one third of patients presented [16]. Atresia of external auditory canals malformation of the middle ear ossicles that may result in bilateral conductive hearing loss [17].
- In the presented case the ears seems normal with normal hearing functions.
- Eyes: Teber *et al*. (2004) mentioned lateral downward sloping of the palpebral fissures (89% of cases) [18].

Sparse or partially absent eyelashes

- A notch in the lower eyelids that is called a coloboma (69% of cases) [11].
- Some people have additional eye abnormalities that can lead to vision loss.
- In the presented case the patient shows all the ophthalmic features and she has defect in cornea formation that leads to loss of vision in one side.
- Other Manifestations: may affects the heart, liver, kidneys, vertebral column and limbs and the brain [16], in the presented case the patient has cerebral atrophy and bowlegs.
- The syndrome may also cause delay in motor and speech development of the affected children.

The diagnosis of this syndrome should be made on 3 basis: Medical history, genetic test and radiographic examination including (CT scan for the patients head to detect bone defects in the ears, zygomatic complex and mandible, Orthopantomogram needed to detect any impacted permanent teeth and to evaluate the condition of the presented teeth).

Other syndromes may have phenotypic overlap with Treacher Collins syndrome, such as Nager syndrome and Miller syndrome. they may have similar clinical features however, in Nager syndrome the characteristic eye feature (lower eye lid coloboma) is absent although the other eye features are similar to those of TCS, also the mandible is more hypoplastic in Nager Syndrome [19]. Miller syndrome also has the same eye features of TCS but cleft palate is more common in TCS, also in Miller syndrome the patient usually has shortening of radius and ulna and congenital heart defects [20].

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