

## A description of Treacher Collins Syndrome in a Yemeni child. A case report

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### Key words

Treacher Collins Syndrome.  
Mandibulofacial dystosis.  
Hereditary diseases.

### Abstract

This article reports a mild case of Treacher Collins syndrome (TCS) in a ten years old child coming to the dental clinic in Ibb University from a distant rural area in Yemen. We described thoroughly the signs & symptoms of the case and proposed a treatment plane that we aimed to be followed .The purpose of this paper is to give informations about this malady because they are absent in medical literatures of this country and to bring medical staff attention to this rare condition.

### Introduction

Edward Treacher Collins, a British Ophthalmologist was first who describe this birth defect(1) . Treacher Collins syndrome (TCS) ) is also called:

- Franceschetti-Zwahlen-Klein syndrome
- Mandibulofacial dysostosis (MFD1)
- Treacher Collins-Franceschetti syndrome
- Zygoauromandibular dysplasia (2).

TCS is a genetic disorder that is passed down in families( hereditary) causing developmental anomalies in structures developing from 1st and 2nd branchial arches , that means ; jaw bones, ears , eye bones and lashes besides palatal and /or lip clefts(3). The syndrome appears to be transmitted in an autosomal dominant manner (i.e. it may be inherited from a parent affected with Treacher Collins with 50% chance of passing it on) with incomplete penetrance and variable expressivity,therefore the condition may vary in severity from generation to generation and from person to person (4) . It may also occur in children of unaffected parents. The chances of Treacher Collins occurring again in children of unaffected parents are minute;

however, new genetic studies could change this opinion (5). More than half of all cases are thought to be due to new gene changes (mutations) because there is no family history of the disease, these mutations in the TCOF1 gene on chromosome 5,cause (TCS) which produces deficiency in protein called treacle that is believed to have a role in facial bones development before birth (6). This condition affects an estimated 1 in 50,000 (7) or 1:10,000 to 1:25,000 (8) of living births. The signs and symptoms of this disorder vary greatly, ranging from almost unnoticeable to severe (9) . People with (TCS) usually have normal intelligence and most children get benefit from early intervention speech and language programs, as well as surgical and orthodontic dental therapies (1,9,10).

### Case description

Ten years old boy was diagnosed at the faculty of dentistry in Ibb University was complaining from pain in his primary lower 2<sup>nd</sup> molar. Family history revealed that the boy was the only affected in his family and his parents were relatives. The boy was from a low economical Yemeni rural society.

Medical history clarified that the boy

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was thin, skinny and appeared to be younger than his age. No systemic abnormalities were detected in his peripheries. His face was calm and funny (figure1) but he was quite intelligent, he was attending regularly a primary school in his village. However we discovered that he was complaining from a recurrent ear troubles with some hearing impairment along his life.

Extra oral examination showed symmetrically elongated face with eyes slanting downwards, wide nose bridge, underdeveloped malar bones making the midface flat (figure 2). The premaxilla was pronouncingly protruded. The lower jaw was retruded obviously and the chin appeared small and tapered (figure 3).

Oral examination showed a relatively large mouth with mixed dentition. Upper centrals were large in comparison with the jaws (figure 4). The boy had a constricted high palate and class II malocclusion with anterior open bite, a small and tapered tongue. The lower jaw was small and the teeth were relatively crowding, the primary molars were decayed.

On the bases of these clinical findings the child was diagnosed to have a mild type of TCS and the following treatment plan was prepared:

- Treatments of the chief complain. Together with insurance to the child's relative about the good prognosis of the treatment in case of following the steps of the plan regularly.
- Oral rehabilitation and motivation of the child for good oral health, beside the motivation of his relative to be aware about the boy condition.
- A programmed scheduled for conservative teeth therapy.
- Thereafter regular continuous visits for orthodontic treatment while the child was growing.
- Continuous and regular visits to the ENT specialist.

Unfortunately, we missed the patient after the completion of the conservative therapy.

## Discussion

TCS is a genetic disorder affecting symmetrically the whole facial region including eye bones, zygomas, upper and lower jaws, chin and ears. That means all structures that have a neural crest origin (1,3,11). Severity differs but all TCS conditions have the same typical facial antimongoloid features ( bird face or fish face like appearance) and some type of ear problems which are variable in severity from absence or deficient external ear to complete deafness(9,12,15,16). The case was clinically diagnosed as TCS depending on the typical and classical signs and symptoms found on examination with what has been cited in the published literatures (12,13,14,15).The child had bilateral symmetrical deformity and did not show any systemic malformations in his body besides ,he was intelligent which leads us to exclude other similar syndromes (14, 17,18). Depending on the ant's tail, our patient was the only affected child in his family which was consisted of other three brothers, this may suggest mutation rather than hereditary type (5).

Some of the living births with TCS can pass without severe or noticeable health complications like in our case (19). Facial deformities in the mild cases can be repaired in several cosmetic and orthodontic options (1,10,20).However we missed our patient ,he discontinued the treatment program after conservative therapy had completed. We think that the negligence of such families to complete treatment is principally because of the low social and economic levels in the rural areas of Yemen and lack of simple health education programs. Moreover, lack of health centers in distant villages and if they are present the health care workers are not aware to these rare conditions.

It is important to motivate physicians and more specifically dentists to be warrant to such rare cases knowing that the congenital hereditary malformations in Yemeni rural areas have a high prevalence percentages because Yemen is considered one of the least developed countries where numerous epidemic and inherited diseases spread, mainly because

of the lack of medical services provided to citizens (21,22).

### Conclusion

Mild cases of TCS can pass peacefully not noticed either from the family or from the health staff. If the condition was not carefully investigated and diagnosed early in life might lead to more serious dental problems.

### Recommendations

- We have to increase social and health awareness among rural societies in Yemen to medical disorders.
- Bring health staff interest to these rare inherited cases in convergent families.
- Encourage researches on hereditary disorders in Yemen.



Figure (1): A calm funny face.



Figure (2): An elongated flat mid face.



Figure (3): Protruded premaxilla & small chin.



Figure (4): Large upper central incisors.

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