

Treacher Collins Syndrome: A Case Report and a Brief Review on Diagnostic Aids

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Case Report

20-year-old boy came to the clinical with chief complaint of multiple carious and malaligned teeth. the patient was examined, he revealed:

downward slanting of eyes, depressed zygomatic arches, sunken cheek bones, deformed external ears, congenital upper eyelid coloboma, retruded chin giving bird-like appearance.

Intraoral examination revealed:

crowding of upper and lower anterior teeth, multiple caries teeth.

The patient also had high arched palate, anterior open bite and bifid uvula.



Introduction

Treacher Collins syndrome is a condition that affects the development of bones and other tissues of the face. The signs and symptoms of this disorder vary greatly, ranging from almost unnoticeable to severe. it caused by abnormal development of the facial 1st and 2nd pharyngeal arches its an autosomal dominant genetic disorder which is caused by mutation in chromosome 5 {5q32-33 } in gene TCOF1 called treacle.

Dental anomilies

- 1. Tooth agensis
- 2. Mandibular second premolars defects
- 3. Enamel opacities
- 4. Cleft palate
- 5. Ectopic eruption
- 6. Bifid uvula
- 7. Malocclusion

Hypodontia

Is an inherited condition characterized by developmentally missing teeth, although absent third molars are a "normal" variation and may not be considered to be part of hypodontia. It is very rare in the primary dentition. Less than six missing teeth have rather small or very conical teeth.



Figure (1)

Mandibular second premolar defect

One of eight bicuspid teeth located in pairs on each side of the upper and lower jaws behind the canines and in front of the molars.



Figure (2)

Enamel opacities

A visibly lighter area on a tooth's surface; maybe caused by fluorosis, or demineralization in high optimum and low fluoride areas have been reviewed in figure 3



Figure (3)

A cleft palate (lip)

Is when the roof of the mouth contains an opening into the nose. These disorders can result in feeding problems, speech problems, hearing problems.



Figure (4)

Bifid uvula

It is often considered as a marker for sub mucous cleft palate. Compared to the normal one, it has fewer amounts of muscular tissues. It is commonly noticed in infants and is rarely found in adults .



Figure (5)

Ectopic eruption

Condition in which the permanent teeth, because of deficiency of growth in the jaw or segment of jaw, assume a path of eruption that intercepts a primary tooth, causes its premature loss and produces a consequent malposition of the permanent tooth.



Figure (6)

Malocclusion

Refers to the alignment of your teeth. In some cases, malocclusion is minor and only causes cosmetic issues.



Figure (7)

Treatment

Orthodontics can help establish to improve the bite and intercuspation, however extraction and implant maybe necessary for some case as repeated surgical intervention combined with poor bone stock the tooth roots to potential devascularisation and injury.

Palatoplasty Is a surgical procedure used to correct or reconstruct the palate in a person with a cleft palate.

conclusion

Management of TCS needs a multidisciplinary approach. The treatment plan is made to meet the individual patients need, considering the growth patterns, function and psychological development. our responsibility to identify this craniofacial disorder, to provide appropriate maxillofacial and orthodontic care, referrals to a geneticist for counselling.

References:

- Dentistry and Scully, C. (2014). Scully's Medical Problems in Dentistry
- Ncbi.nlm.nih.gov. (2018). *Home PMC NCBI*
- Sciencedirect.com. (2018). ScienceDirect.com | Science, health and medical journals, full text articles and books.