Review Article

Mandibulofacial Dysostosis: A Case Report

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ABSTRACT

Mandibulofacial dysostosis is also known as Treacher Collins syndrome (TCS) or Treacher Collins–Franceschetti syndrome is a rare autosomal dominant congenital disorder characterized by craniofacial deformities, such as absent cheekbones. Treacher Collins syndrome is found in about 1 in 50,000 births. The typical physical features include downward slanting eyes, micrognathia (a small lower jaw), conductive hearing loss, underdeveloped zygoma, drooping part of the lateral lower eyelids, and malformed or absent ears.

Keywords: mandibulofacial dysostosis, treacher Collin syndrome.

INTRODUCTION

Named after Edward Treacher Collins (1862 1932)the English surgeon and ophthalmologist who described its essential traits in 1900.[1] In 1949, Adolphe Franceschetti and David Klein described the same condition on their own observations as mandibulofacial dysostosis. The term mandibulofacial dysostosis is used to describe the clinical features.[2] Mutations in the TCOF1, POLR1C, or POLR1D gene can cause Treacher Collins syndrome.

CASE REPORT

A 15 year old female patient visited the OMR department of Jaipur Dental College with the chief complain of malalignment of teeth. On extraoral examination, ocular hypertelorism, auricular deformity, mandibular hypoplasia, brachycephalic face was noticed while intraorally, high arched palate and enamel hypoplasia was present.

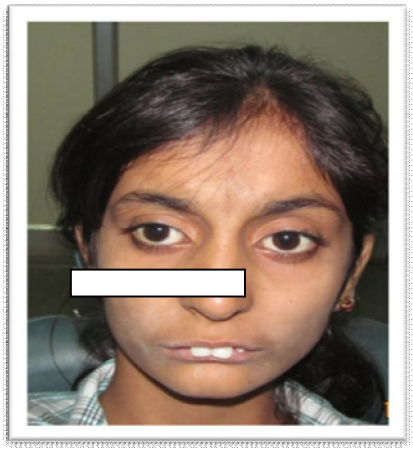


Fig. 1 shows ocular h:pertelorism, auricular deformity, mandibular hypoplasia, brachycephalic face

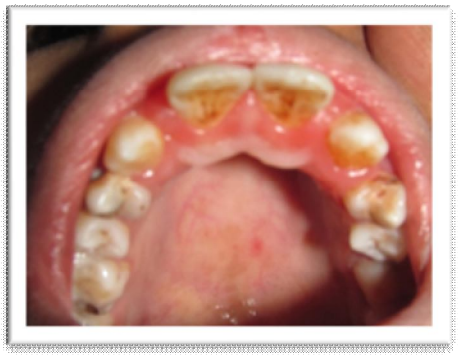
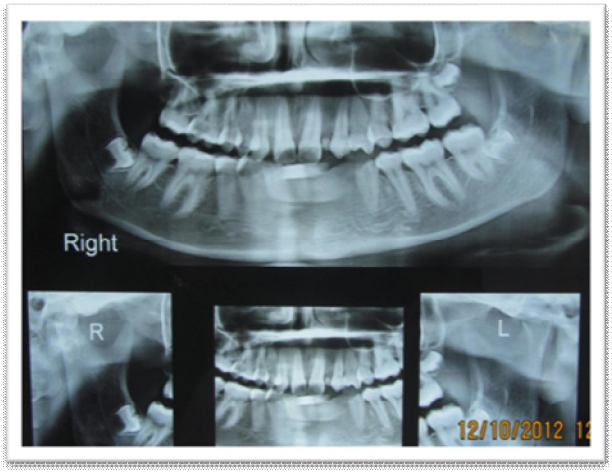


Fig. 2: shows high arched palate

Hence, based on clinical features provisional diagnosis of Treacher Collin Syndrome was given. For further confirmation an OPG was advised.



OPG reveals hypoplasia of mandible. Therefore, finally Treacher Collin Syndrome was diagnosed.

DISCUSSION

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. Most affected individuals have underdeveloped facial bones, particularly the cheek bones, and a very small jaw and chin (micrognathia).[3] People with Treacher Collins syndrome often have eyes that slant downward, sparse eyelashes, and a notch in the lower eyelids called an eyelid coloboma. Some affected individuals have additional eye abnormalities that can lead to vision loss. This condition is also characterized by absent, small, or unusually formed ears. Hearing loss occurs in about half of all affected individuals; hearing loss is caused by defects of the three small bones in the middle ear, which transmit sound, or by underdevelopment of the ear canal. Patients with Treacher Collins syndrome usually have normal intelligence.[4] Our patient was also mentally normal. Condition affects an estimated 1 in 50,000 people. Treatment of mandibulofacial dysostosis (Treacher Collins syndrome) is lengthy and requires a multidisciplinary approach focused on treatment of symptoms.[6]In newborns with mandibulofacial dysostosis, immediate attention to airway and swallowing inadequacies is critical.[7] Family-to-family support has proven to be of great psychological value. Operative repair of Treacher Collins syndrome is based upon the anatomic deformity and timing of correction is done according to physiologic need and development. Drug therapy is not currently a component of the standard of care for this syndrome.In our patient only symptomatic relief was given as replacement of missing teeth along with orthodontic treatment.

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