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Case Report

Treacher Collins Syndrome-A Mild Variant

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Abstract: We report a case of a 12 year old female patient with Treacher Collins Syndrome affecting only the eyes and the jaw. The diagnosis of Treacher Collins syndrome was made based on clinical and radiographic findings.
Keywords: Treacher Collins Syndrome, Mandibulofacial Dysostosis, coloboma, anti-mongoloid palpebrae, autosomal dominant.

INTRODUCTION

Treacher Collins syndrome (TCS) or Mandibulofacial Dysostosis (MFD) is an autosomal dominant disorder characterized by unusual clinical features associated with abnormalities of structures derived from first and second branchial arches, resulting in defective orofacial features[1-2]. The essential features of this syndrome were described by Treacher Collins in the year 1900, but the first extensive description of the condition was described by Franceschetti and Klein in 1949, and they used the term Mandibulofacial Dysostosis[3].

CASE REPORT

A 12 year old female patient came to the department with complaint of malaligned teeth. On examination we noticed hypoplastic zygomatic complex (Figure 1), anti-mongoloid slanting of the palpebral fissures, colobomata of the lower eyelids, drooping of the lateral part of lower eyelids, a paucity of lid lashes medial to the defect, and hypertelorism (Figure 2). The upper lip was hypotonic and the lower jaw was deviated to the right side. (Figure 1) Intra-oral examination showed, narrow-high arched palate, missing upper laterals and canines. (Figure 3) The patient presented with class II division I malocclusion and convex profile.

On radiological investigations, orthopantamograph showed, horizontally impacted canine on right side, erupting left canine. There was partial anodontia of lateral incisors on either side (Figure 4). The diagnosis was based mainly on clinical and radiological features.

Fig-1: showing hypoplastic zygomatic complex, hypotonic upper lip and lower jaw deviated to right side.

Fig-2: Showing anti-mongoloid slanting of the palpebral fissures, colobomata of the lower eyelids, drooping part of the lateral lower eyelids, a paucity of lid lashes medial to the defect, and hypertelorism.

Fig-3: Showing narrow-high arched palate, missing upper laterals and canines.

Fig-4: Orthopantamograph showing, horizontally impacted maxillary canine on right side, erupting maxillary left canine. There was partial anodontia of maxillary lateral incisors on either side.

DISCUSSION

TCS is a rare genetic disorder characterized primarily by abnormalities in the development of the head and face. The disorder is characterized by hypoplastic facial bones, particularly the jaws and zygomatic complex, abnormalities of the pinnae that are frequently associated with atresia of the external auditory canals along with anomalies of the middle ear ossicles causing bilateral conductive hearing loss[4,5]. Along with these characteristics other features like anti-mongoloid slanting of the palpebral fissures, colobomata of the lower eyelids, a paucity of lid lashes medial to the defect, Cleft palate are commonly seen. These clinical features are usually bilaterally symmetrical. Our patient presented only fewer features which typically involve the eyes and the jaws.

TCS or MFD is an autosomal dominant disorder with high penetrance and variable expressivity[6]. Other modes of inheritance such as autosomal recessive transmission and a role for gonadal mosaicism and chromosomal rearrangement in the causation of this syndrome have also been proposed[7]. While 40% of cases have a previous family history, the remaining 60% appear to arise as a result of a de novo mutation[7]. This can create an additional complication in providing genetic counseling where the diagnosis in either of an affected child's parents is in doubt. Our patient did not yield any relevant family history.

The treatment of TCS cases depends on the severity. A multidisciplinary craniofacial team approach is critical to coordinate oral, ocular, dental, pediatric, and craniofacial care. Neonatal mandible advancement is required to relieve airway obstruction secondary to characteristic microretrognathic TCS mandible. Surgical intervention is made to relieve skeletal malooclusion, followed by orthodontic treatment to correct bite and intercuspation. Palatoplasty is performed in the presence of cleft palate. Finally Soft tissue reconstruction for ocular and auricular defects is required. Since our patient did not have any major functional liabilities she was referred for Orthodontic correction of malocclusion and Prosthodontic rehabilitation of missing teeth.

CONCLUSION

The Management of TCS includes genetic consultation, evaluation, and treatment planning to be provided by an experienced multidisciplinary staff composed of representatives from a variety of medical, dental, and other health care specialties. Newer genetic studies need to be carried out, along with pre-natal counselling to aid in diagnosis and also for better management of TCS patients

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