Cleidocranial Dysplasia - A Rare Case Report.

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ABSTRACT

Cleidocranial dysplasia is an autosomal dominant heritable disorder caused by mutation in the gene encoding transcription factor Core Binding Factor Subunit Alpha 1 (CBFA1) or Runt related transcription factor 2 (RUNX2). It is characterized by aplasia or hypoplasia of the clavicles, characteristic craniofacial malformations, and the presence of numerous supernumerary and unerupted teeth. A 37 year old male with cleidocranial dysplasia is described in this article with cyst associated with impacted teeth as the first presentation.

Keywords: Autosomal Dominant, Mutation, Cleidocranial dysplasia.

INTRODUCTION

Cleidocranial dysplasia(CCD) is an autosomal dominant heritable disease which is characterized by hypoplasia or aplasia of the clavicle, abnormal growth of facial bone, and relatively rare skeletal and dental developmental abnormalities with delayed eruption or impaction of teeth.[1] It is also known as Marie and Sainton's disease, mutational dysostosis or Cleidocranial dysostosis . The first case of clavicular defects was reported by Martin in 1765. Cleidocranial dysplasia was first described by Pierre Marie and Paul Sainton in 1898. It is caused by mutation in the gene on 6p21 encoding transcription factor Core Binding Factor Subunit Alpha 1 (CBFA1) or Runt related transcription factor 2 (RUNX2). RUNX2 is a part of the Fibroblast Growth Factor (FGF) and Bone Morphogenetic Protein (BMP) signaling pathways in tooth and bone development respectively.[2]

CASE REPORT

A 37- year- old male patient presented to the Department of Oral and Maxillofacial Surgery, Government Dental College and Hospital Srinagar, Jammu and Kashmir with a chief complaint of swelling on left side of lower jaw since 1 year. Past medical and dental history was insignificant. General physical examination revealed short stature, ability to approximate his shoulders anteriorly. He had prominent forehead with frontal bossing, mid-facial hypoplasia with depressed nasal bridge and hypertelorism. Intraorally, high arched palate, narrow maxilla, multiple missing teeth in both maxillary and mandibular arch. A panoramic radiograph was taken and it indicated multiple unerupted permanent teeth and supernumerary teeth, a narrow ascending ramus, cyst formation with supernumerary teeth in premolar region. Chest radiograph revealed narrow thorax, absent clavicles. PA view skull revealed incomplete closure of interparietal suture [Figure 1-7]. Biochemical investigations showed normal serum calcium and phosphate level. All investigations and radiograph were confirmatory for the diagnosis of CCD.

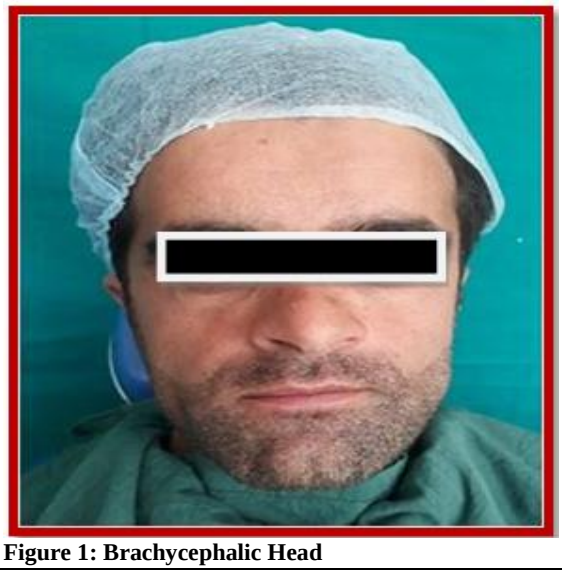

Figure 1: Brachycephalic Head

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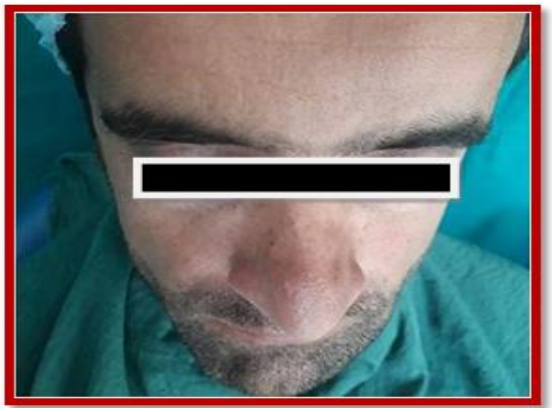


Figure 2: Depressed Nasal Bridge

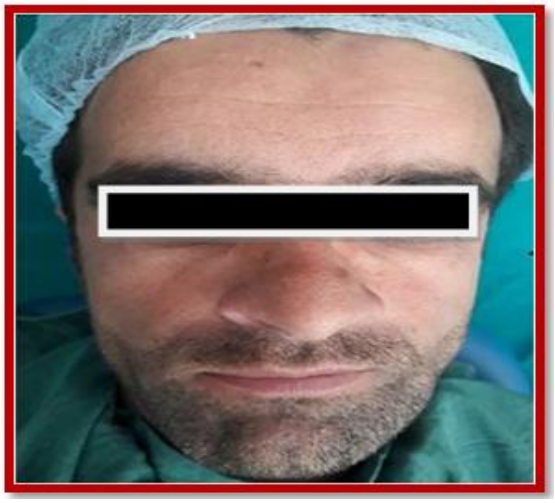


Figure 3: Frontal Bossing and Hypoplastic Maxilla Maxillaandhypoplastic Middle

Figure 4: Absence of Clavicles

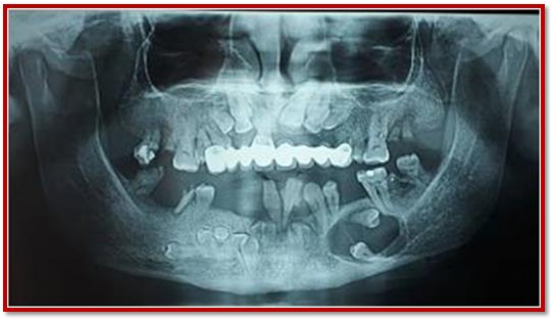


Figure 5: OPG Showing Multiple Impacted Teeth with Associated Cyst

Figure 6: Absent Clavicles

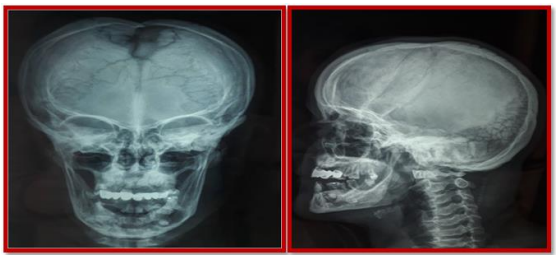
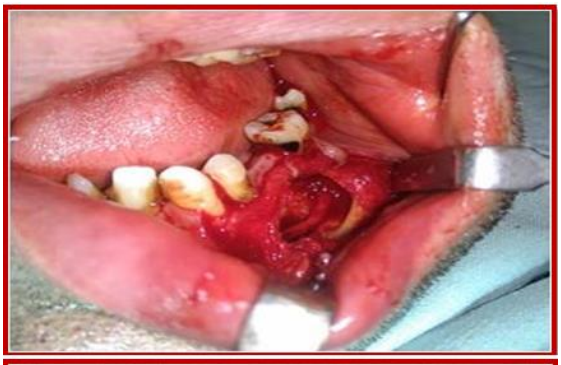


Figure 7: Widened Sutures with Wormian Bone



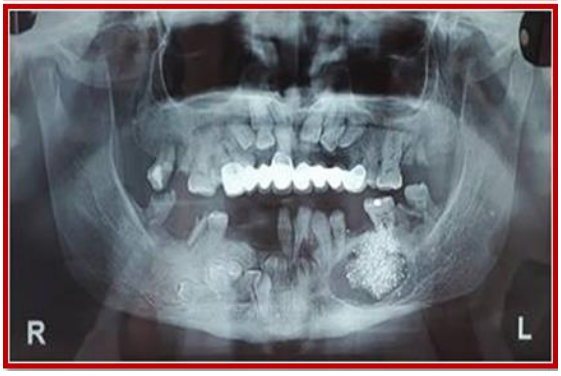


Figure 8-9: Enucleation of Cyst Along With Removal of Impacted Teeth Followed By Secondary Packing With Iodoform Dressings.

Treatment Plan

A cardiology and general physician checkup was done for the patient which did not reveal any major abnormality at present except slight cardiac anomaly in the form of ST segment elevation .Patients with CCD have a variety of structural abnormalities that may potentially interfere with patency of the airway requiring careful assessment

by the anesthesiologist involved in their care. Anatomical abnormalities of the skull and the facial structures (including dentition) may impede with mask ventilation and endotracheal intubation while spinal abnormalities may pose challenges to neuraxial techniques. After thorough evaluation patient was cleared for surgery under local anesthesia. Eucleation was done along with removal of impacted teeth [Figure 8-9]. Patient was treated with regular follow ups.

DISCUSSION

This rare hereditary skeletal disorder, which is also known as Scheuthauer-Marie-Sainton syndrome or Cleidocranial dysostosis, is characterized by abnormal skeletal and dental development. The prevalence of CCD is an estimated one per million and does not differ by race or by gender. In most cases, the disorder is an inherited autosomal dominant trait. In 20 -40% of reported cases, however, the disorder occurs sporadically.[3] The etiology though not completely known is thought to be due to a CBFA1 (core binding factor activity 1) gene defect on the short arm of chromosome 6p21. CBFA1 is essential for differentiation of stem cells into osteoblasts, so any defect in this gene will cause defects in the membraneous and endochonral bone formation.[4] It was first reported in 1760 and until now, more than 1000 cases of CCD have been reported in medical literature.[5] In general, CCD patients are of a normal intelligence quotient and health. They are slightly dwarfed and slender and have long necks and drooped shoulders.[6] The most striking skeletal defects are hypoplastic or aplastic clavicles, late closure of the fontanelles, open skull sutures and multiple wormian bones. Partial hypoplasia commonly involves the acromial end of the clavicles. Complete absence of clavicles is seen in 10% of cases]. The thoracic cage is small and bell or cone shaped with short, oblique ribs making the individual prone to chest infections.[2] Other features include brachycephaly, prominent forehead, depressed nasal bridge, hypertelorism, midfacial hypoplasia, narrow high arched palate, conductive deafness, scoliosis,respiratory distress ,recurrent sinus abnormalities.[7] Dental abnormalities are typical main features of CCD, and they occur in 93.5% of affected patients. The primary dentition usually develops relatively normally, while the permanent dentition is severely disturbed. Supernumerary teeth, prolonged retention of the primary dentition, failed eruption of the permanent teeth, multiple crown and root abnormalities are the more common dentition abnormalities.[8] The signs and symptoms described here should serve as early markers to aid the general and pediatric dentist in planning appropriate treatment or referring patients to specialized centers. An interdisciplinary treatment approach involving orthodontics, maxillofacial surgery and prosthodontics is obligatory. In confirmed cases, genetic counseling for family planning should certainly be advised.[6]

CONCLUSION

Although Cleidocranial Dysplasia is not encountered routinely in dental practice, early diagnosis through oral finding is possible.In addition to oral features diagnosis of this rare syndrome requires a reliable skeletal evaluation. Family history, pathognomonic clinical and radiographic findings play a central role in the diagnosis of CCD. Treatment of these patients requires a multidisciplinary approach.

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