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CLEIODOCRANIAL DYSPLASIA – A RARE CASE REPORT

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Conflicts of Interest: Nil

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Abstract:

Background and aim: Cleidocranial dysplasia(CCD) also known as cleidocranial dysostosis is an autosomal dominant malformation syndrome. It affects both sexes equally. Its prevelance is estimated 1:1 million. It can be herited or arises as result of sporadic mutation. Cleidocranial dysplasia is caused by a mutation in the RUNX2 gene on chromosome 6. The most prevalent features associated with CCD are aplastic or hypoplastic clavicles, impacted supernumerary teeth, delayed eruption of permanent teeth, and a hypoplastic maxilla. Hence,this case report was presented with the aim to show the salient features of cleidocranial dysplasia.

Case presentation: We hereby report a case of cleidocranial dysplasia in a 16 year old male patient with no familial pattern of inheritance since the patient is the only member in the family suffering from such disorder. We reported this case because of the extremely low incidence of this disorder.

Conclusion: Early diagnosis allows a proper orientation for the treatment, offering a better compliance to the patient and with anticipatory guidance people with CCD lead healthy and productive life.

Keywords: cleidocranial dysplasia, delayed eruption, autosomal dominant, mutation

Background:

Cleidocranial dysplasia (CCD) also known as Marie and Saiton disease, Scheuthauer- Marie-Sainton syndrome, Mutational dysostosis.[1] CCD is a rare disease with autosomal dominant inheritance characterized by clavicular aplasia or hypoplasia, retarded cranial ossification, impacted supernumerary teeth, relative short stature and a variety of other skeletal abnormalities.[2] This condition is of clinical significance to the dentist due to involvement of facial bones, delayed eruption patterns and presence of multiple impacted supernumerary teeth.[3] CCD was first described by Pierre Marie and Paul Sainton in 1894.[4] The significant features for diagnosis of CCD are multiple supernumerary teeth, partial or complete absence of clavicle and open fontanelle.[5] The aim of this article is to illustrate the clinical features, radiological features and dental abnormalities in a rare case of cleidocranial dysplasia.

Case report

A 16-year-old male patient reported to the Department of Oral Medicine and Radiology DJ College of Dental Sciences and research with a chief complaint of missing upper front tooth and prolonged retention of deciduous teeth. The family history was not contributory. Upon general examination, he was of relative short stature and his gait was normal. There was frontal and parietal bossing and no hypertelorism. The patient has large brachycephalic head, angular face and drooping shoulder with hypoplasia of clavicle. The midface of the patient was depressed due to underdeveloped maxilla and relative prognathic mandible. (Fig.1)

On intraoral examination revealed the presence of retained deciduous and permenant teeth viz.

17,16,55,14,12,63,24,65,26,27,31,35,36,37,41,83,84, 85,46 of which 55, 84, and 85 are decayed. (Fig.2)

Further on radiographic examination, the orthopantomogram outlined the presence of total 42

teeth in both the jaws. Out of these, 19 were seen in oral cavity and 23 were impacted. The mandibular notch showed excessive deepening causing the elongated appearance of coronoid process. (Fig.3)

In Lateral skull view depression of anterior fontanel and prominent wormian bones in the lamboid suture region. Posterior fontanel region was also open as seen in radiograph. The hypoplastic appearance of maxilla was clearly evidenced on the lateral skull radiograph.(Fig.4)

Chest x ray revealed the presence of hypoplastic clavicles. (Fig.5)

Based on clinical, radiological and laboratory findings, the patient was confirmed with cleidocranial dysplasia. He is currently being treated with a multi-disciplinary approach by a team comprising oral physicians, pedodontists, orthodontists and pediatricians, giving prime consideration to growth and development of the facial structures, along with psychological support. His grossly decayed retained deciduous teeth have been extracted and are provided with space maintainers.

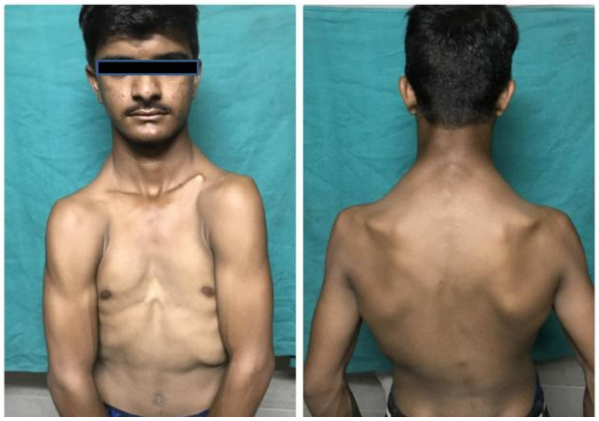

Figure 1: Facial view showing a brachycephalic head and hypermobility of the shoulders.

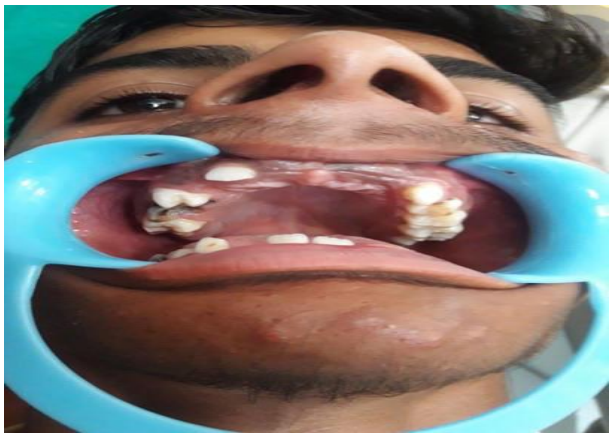

Figure 2: Intraoral view showing persistence of primary dentition.

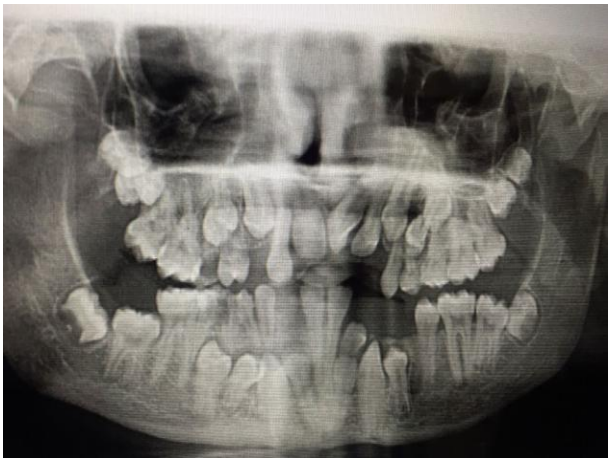

Figure 3: Panoramic X-ray showing supernumerary teeth and impaction.

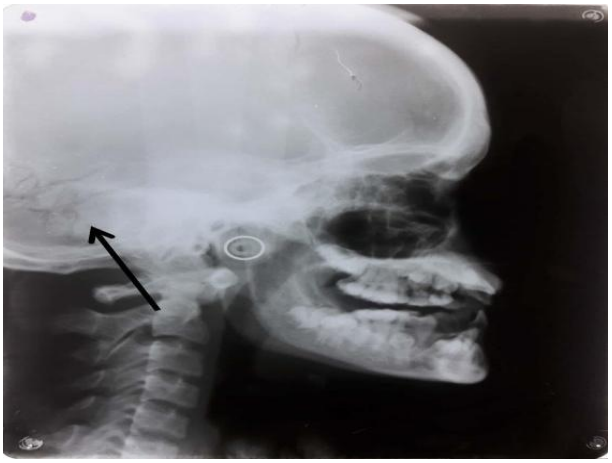

Figure 4: Lateral cephalometric X-ray showing numerous wormian bones, large fontanelles, hypoplastic maxilla.

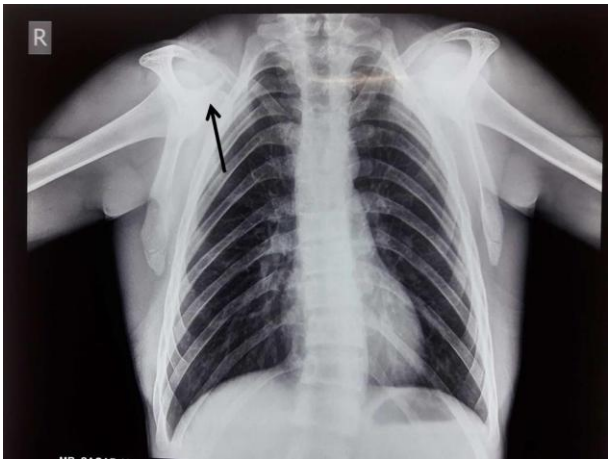

Figure 5: Chest radiograph showing hypoplastic clavicles.

Discussion:

CCD is an autosomal dominant condition characterized by generalized dysplasia of the bones and teeth. The more significant features of the defect in the clavicle and cranium made Marie and Sainton to coin the term cleidocranial dysostosis for this

condition. However, the more generalized dysplasia of bones and teeth has led to the abandonment of "dysostosis" in favour of "dysplasia". The prevalence of this disorder is one per million individuals. The CCD gene is located on either the long or short arm of chromosome 6p21 .[6,7] Zheng et al. reported that humans with CCD have altered endochondrial ossification due to perturbed RUNX2 regulation of hypertrophic chondrocytes[8]. This gene is very essential for osteoblast and dental cell differentiation, and thus for normal bone and tooth formation.

A familial incidence was recorded in approximately 2/3 rd of the reported cases of CCD and the condition was found in as many as five successive generations. When inherited, it appears as an autosomal dominant disease.[9] In those cases which appeared to have developed sporadically, as with the case presented here, it has been suggested that they represent a recessively inherited disease or more likely either an incomplete penetrance in a genetic trait with variable gene expression or a true new dominant mutation.[10] In our case, the patient did not report the existence of direct ancestors or descendents who presented any clinical characteristic of CCD.

Patients usually have mild disproportionate short stature with short limbs compared to trunk, more apparent in upper limbs than in lower limbs. Though females showed more prevalence than male patients.[11] But in our study male was affected.

Some reported cases of CCD have shown biochemical signs of hypophosphatasia including decreased levels of serum alkaline phosphatase.[1] Our patient showed normal alkaline phosphatase, calcium levels and phosphorus level.

The desired treatment of patients with CCD requires a team approach and a compliant patient. The multidisciplinary treatment approach involving orthodontics, maxillofacial surgery, and prosthodontics is compulsory. Good alliance among the specialists, the patient, and the patient's family is essential for an organized treatment approach in which each member can contribute his or her expertise for the best treatment outcome. The main treatment goal is to establish functional occlusion and an aesthetic facial and dental appearance.

Conclusion:

CCD is very rarely encountered in clinical practice and is generally diagnosed accidentally. The clinician should be aware of the characteristic features of CCD for early diagnosis and initiating the appropriate treatment approach. This patient refused an elaborate surgical correction. Retained deciduous teeth are extracted and space maintainers are given to patient.

Clinical significance: CCD is very rarely encountered in clinical practice and is generally diagnosed accidentally. The clinician should be aware of the characteristic features of CCD for early diagnosis and initiating the appropriate treatment approach.

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