

Cleidocranial dysplasia – A case report

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Abstract

Cleidocranial dysplasia is an autosomal dominant disorder characterized by bony dysplasia in clavicles, patent sutures and fontanelles, short stature and multiple unerupted teeth. Despite its variable expressivity, early diagnosis through the classical findings can be achieved by clinicians. The diagnosis of this disease requires a reliable skeletal evaluation in addition to the oral findings. Radiographs of the skull and chest are important to diagnosis of Cleidocranial dysplasia. A case report of 11yrs boy who manifested with classical features of this disorder is presented here.

Keywords: Cleidocranial dysplasia, Delayed eruption, Retained teeth, Impacted teeth.

Introduction

Cleidocranial dysplasia (CCD) is an autosomal dominant disorder characterized by skeletal dysplasia in clavicles, patent sutures and fontanelles, the formation of wormian bones and short stature. Mutations of Runx2 gene on chromosome 6p21 has been identified as being responsible for cleidocranial dysplasia.¹ In this article, we present a case report of a child with classical features of CCD.

Case presentation

A 11yrs boy reported with the complaint of retained deciduous teeth. Patient's deciduous teeth had erupted at normal age, but had not exfoliated. Past dental history, medical history and family history were non contributory. Physical examination revealed relative short stature & hyper mobility of shoulder joints. Patient could bring his both shoulders to midline (Fig. 1). Facial features revealed brachycephalic skull. Soft bone could be felt at lambda region. Face was triangular shaped with broad forehead, depressed nasal bridge, deficient malar eminence and straight profile.

Intra oral Examination revealed deep palatal vault. All deciduous teeth and permanent first molars had erupted (Fig. 2 & 3). A panoramic radiograph showed multiple impacted permanent teeth (Fig. 4). There were no supernumerary tooth buds present. Lateral cephalogram revealed hypoplastic maxilla & relatively prognathic mandible (Fig. 5). Posteroanterior skull radiograph revealed bulb shaped skull with metopic suture and patent anterior fontanelle (Fig. 6). Chest radiograph showed complete absence of clavicles, bell shaped chest & spina bifida occulta (Fig. 7). The case was diagnosed as Cleidocranial dysplasia. The patient was further evaluated by pedodontists and was planned for arch expansion followed by orthodontic treatment.

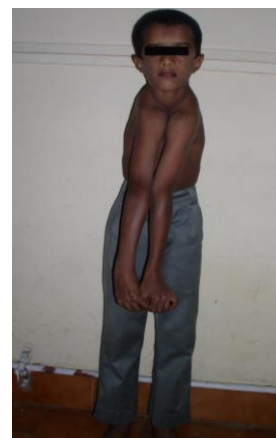


Fig.1: Approximation of both the shoulders at midline



Fig. 2 & 3: Intraoral photograph depicting the dentition



Fig. 4: Panoramic radiograph showing multiple impacted permanent teeth



Fig. 5: Lateral cephalogram revealing hypoplastic maxilla & relatively prognathic mandible



Fig. 6: Posteroanterior skull radiograph showing bulb shaped skull with open sutures



Fig. 7: Chest radiograph showing complete absence of clavicles & spina bifida occulta (inset)

Discussion

Marie and Sainto were the pioneers to describe the congenitally missing clavicles, a characteristic manifestation of CCD, in 1897. Mutations of Runx2 gene [also known as Core-binding factor $\alpha 1$ (*CBFA1*)] on chromosome 6p21 have been identified as being responsible for CCD. Previous researches have shown that mutation is a submicroscopic deletion at 6p21. This gene encodes a transcription factor which has the DNA binding ability in the Runt-domain. *CBFA1* controls osteoblast differentiation from precursor cells and is essential for bone formation.^{2,3}

CCD shows a variable somatic presentation. The defective ossification of endochondral and intramembranous bones may lead to delayed or failure of ossification of portions of the skeletal structure. Although a classic triad of cranial, clavicular, and pelvic deformities are the most striking changes, Individuals may present with all or some of the characteristic features. The classical aspects of this disorder consist of large brachycephalic head, patent anterior fontanelle, persistent metopic suture, small and angular face, prominent frontal and parietal bones and drooping shoulders with excessive mobility, nasal bone absence, non-union of the mandibular symphysis, bipartite hyoid bone, spina bifida occulta and delayed closure of the symphysis pubis.⁴ Depending on the amount of clavicular involvement, the patient may be able to approximate the two acromial regions below the chin. As the clavicle was completely absent, our patient could bring his both shoulders to midline. Height is reduced in both sexes, but dwarfism is not a common finding.

Because of delayed and disturbed mineralization, there may be abnormal dentition unerupted or late erupted deciduous and permanent teeth. The teeth may appear small, irregularly spaced, or crowded. Impacted multiple supernumerary teeth is a common feature observed in CCD.⁵ although our patient had impacted teeth as evident in panoramic radiograph, there were no supernumerary teeth present.

The most marked and consistent disturbance observed was the extreme delay or arrest of physiologic root resorption and shedding of the teeth. This finding is probably related to the generalized reduced bone resorption observed in the jaws in CCD and to the lack of eruption of the permanent teeth. The hypothesis of incomplete or markedly delayed resorption of the dental lamina is in accordance with previously reported abnormal bone remodelling with diminished bone resorption, seem to contribute to the development of supernumerary teeth in CCD.⁶

Conclusion

Though CCD presents with variable presentations, early diagnosis through the classical findings can be achieved by clinicians. The diagnosis of this disorder involves oral as well as complete skeletal evaluation.

Radiographs of the face and skull are important in the diagnosis of CCD. Dental practitioners play an important role in the diagnosis and management of CCD. It requires careful planning and the collaboration of many dental specialists. An functionally efficient permanent dentition and an aesthetically appealing facial appearance may be achieved by interdisciplinary treatment when CCD is diagnosed in the early stages of development.⁷

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