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## IMAGES IN MEDICINE

### Cleidocranial Dysplasia – Report Of a Case

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#### ABSTRACT

Cleidocranial dysplasia is a rare congenital disorder of the bone, characterized by abnormalities of the clavicles, skull and jaw bones. It was first described by Pierre Marie and Paul Sinton in 1898 and since then, over 1000 cases have been documented in medical reports. It is a rare syndrome which is usually caused by an autosomal dominant gene, although 40% of the cases of CCD appear spontaneously, with no apparent genetic cause. The bony and dental features of CCD may be visualized on the radiographical images of the face and the skull and they are characteristic. We report here, a case of Cleidocranial dysplasia in an 18 year old patient, with emphasis on radiological findings.

**Key Words:** Cleidocranial dysplasia, impacted teeth, Marie Sinton Disease, Clavicle, Mutational dysostosis

**Key Message:** Oral radiologists play an important role in diagnosing systemic diseases which are manifested in the jaws.

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Cleidocranial dysplasia is a well known, rare syndrome which is usually caused by an autosomal dominant gene, usually caused by a mutation of the Core Binding Factor –  $\alpha 1$  gene, located at chromosome 6p21 [1],[2],[3].

CCD is also known as the Marie and Sinton disease, Mutational dysostosis and Cleidocranial dysostosis [4]. The disorder was originally thought to involve the bones of intramembranous origin only, namely the bones of the skull, clavicles and flat bones. However, it is now known that the bones of endochondral ossification are also affected and that it is a generalized disorder of many skeletal structures and therefore, the substituted term ‘cleidocranial dysostosis’ was given [5].

#### Introduction

CCD presents with skeletal defects of several bones, the most striking of which are the partial or complete absence of the clavicles, which allows the patient to approximate the shoulders anteriorly in the midline [6].

The facial abnormalities include underdeveloped paranasal sinuses, ocular hypertelorism, mild exophthalmus and mandibular prognathism leading to midface deficiency. Characteristically, these patients show prolonged retention of deciduous dentition and multiple supernumerary impacted teeth, often mimicking a premolar [5].

### Case Report

An 18 year old male patient reported to our department with a chief complaint of pain in the lower jaw.

The head and neck examination revealed frontal and occipital bossing and the presence of a metopic suture. The clavicles were absent on both the sides and the patient was able to get his shoulders anteriorly in the midline when asked to do so [Table/Fig 1].



(Table/Fig 1) Shoulders Approximating In The Midline Due To Absence Of Bilateral Clavicles.

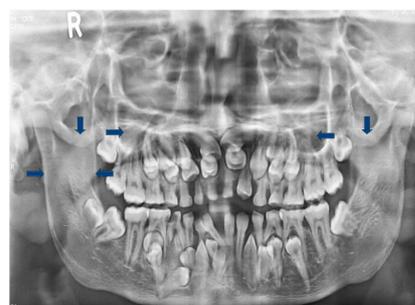
Examination of the oral cavity revealed over retained deciduous teeth and the presence of the first, second permanent molars and lower incisors in the entire oral cavity [Table/Fig 2]. Dental caries was noted in relation to right mandibular 2<sup>nd</sup> molar and the tooth was tender on percussion.



(Table/Fig 2) Oral Cavity Showing Retained Deciduous Teeth.

Based on the clinical findings, the patient was provisionally diagnosed as Cleidocranial dysplasia and was therefore, subjected to routine radiological examination.

The panoramic radiograph of the patient revealed the presence of a total number of 56 teeth, including both erupted and unerupted impacted teeth [Table/Fig 3]. The overall size of the maxillary sinuses was reduced. The ascending ramus of the mandible was parallel sided and the coronoid process was pointed and curved distally. The zygomatic arches on both sides were drooping downwards on the ramus of the mandible.



(Table/Fig 3) OPG Showing Multiple Impacted Teeth, Reduced Size Of Bilateral Maxillary Sinuses, Parallel Shaped Ramus Of Mandible And Zygomatic Arch Being Bent Anteriorly On The Ramus.

The chest radiograph was characteristic in appearance, i.e. a narrow bell shaped thoracic cage, the complete absence of bilateral clavicles, small scapulae and small glenoids and the ribs were directed obliquely downwards [Table/Fig 4]. The skull radiographs revealed open sutures, multiple wormian bones in the occipital region and

sphenoids which showed the hypoplastic presence of a metopic suture [Table/Fig 5], [Table/Fig 6]. The overall shape of the skull was of a 'light bulb', with midface deficiency and mild prognathism of the mandible.



(Table/Fig 4)Chest Radiograph Showing Bilateral Absence Of Clavicles, Bell Shaped Thoracic Cage And Ribs Directed Obliquely Downwards.



(Table/Fig 5)Lateral Skull Showing Multiple Wormian Bones In The Occipital Region.



(Table/Fig 6)AP View Showing Open Sutures And Metopic Suture.

## Discussion

CCD is a rare congenital defect of autosomal dominant inheritance, with skeletal defects of several bones [5]. The clavicles may be rudimentary, or completely absent [6]. Delayed ossification of the cranial sutures and fontanelles, coupled

with raised hydrocephalic pressure results in expansion of the cranial vault, leading to bossing of the frontal, parietal and occipital bones [5]. Similar findings were noted in our case too.

According to Jensen and Kreiborg, the zygomatic bone is hypoplastic with the arch being thin or even discontinuous and having a characteristic downward bend [6].

Characteristically, patients with CCD show prolonged retention of deciduous dentition and delayed eruption of permanent teeth. As many as 63 unerupted teeth have been documented in one patient [5]. The panoramic radiograph of our patient revealed 56 teeth. Jensen and Kreiborg have suggested that supernumerary teeth form as a result of the activation of the remnants of the dental lamina which are left unresorbed during odontogenesis [7].

In terms of the dental management of CCD, several approaches have been reported over the years. The option of no treatment was common in the past. The current "state of the art" management involves a multistaged treatment protocol of the timely extraction of deciduous teeth, staged surgical removal of supernumerary teeth, exposure of selected unerupted permanent teeth and orthodontic forced eruption [8]. Our case has also been referred to the department of Oral and Maxillofacial Surgery and Orthodontics for further needful.

## Conclusion

CCD is a condition which is usually present since birth, but is often missed or diagnosed through incidental findings by the physician, the same as in our case.

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