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Cleidocranial Dysplasia with Normal Clavicles: A Case Report Highlighting Skeletal and Dental Variability

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Abstract: ***Cleidocranial dysplasia (CCD) is a rare skeletal disorder characterized by anomalies in bone and dental development, typically associated with underdeveloped or absent clavicles. This case report presents a 19-year-old male with classic features of CCD, including multiple impacted and supernumerary teeth, open skull sutures, and mid-facial hypoplasia, but notably with normally developed clavicles—a rare variation. Diagnosis was established through clinical, radiological, and CT imaging, followed by surgical intervention involving cyst enucleation and extraction of impacted teeth. This case highlights the phenotypic variability of CCD and underscores the importance of comprehensive evaluation in atypical presentations.***

Keywords: cleidocranial dysplasia, RUNX2 mutation, impacted teeth, skeletal anomalies, dental radiology

1. Introduction

Cleidocranial dysplasia (CCD) is a generalized skeletal abnormality which is typically inherited in an autosomal dominant pattern due to a mutation in the RUNX2 gene (1) It is distinguished by a wide range of common skeletal and clinical abnormalities. Patent cranial sutures and fontanelles, brachydactyly, dental abnormalities, clavicular aplasia/ hypoplasia, pelvic hypoplasia, and short stature are among the skeletal abnormalities common to CCD. (2) Males and females are equally afflicted by the condition, which has an incidence of 0.5/100,000 live births. They typically exhibit small stature and skeletal and dental anomalies. Despite the autosomal dominant nature of CCD inheritance, approximately one-third of affected cases have spontaneous mutations.

Although most cases are considered sporadic, familial clustering has been reported. Mutations in the RUNX2 gene at 6p21.1 in the little arm of chromosome 6 are the causes of CCD.(3) (4)Marie and Sainton first identified the disorder as cleidocranial dysostosis (5). Cleidocranial dysplasia is the name given to this illness because it is now recognized as a common anomaly in bone growth. (6)

The purpose of this case report is to describe an atypical presentation of cleidocranial dysplasia with normally developed clavicles and to emphasize the diagnostic and therapeutic implications of this variation.

2. Case Report

A 19-year-old boy presented to the department of Oral Medicine and radiology with the chief complaint of pain in the right lower back tooth region for the past one month and swelling since yesterday. His complaint was pain that was gradual in onset, dull and throbbing, occurring only during chewing, and self-resolving. He gave the history of swelling in the right lower back tooth region since yesterday which is sudden in onset. The boy was delivered by normal vaginal delivery, no relevant past medical history this was his 1st dental visit.

On general examination his height -137 cm, weight was 31 kg, gait was normal, very thin. On extra oral examination no gross facial asymmetry was noticed, depressed nasal ridge, widely spaced eyes (hypertelurism), hypoplastic maxillae (fig 1).

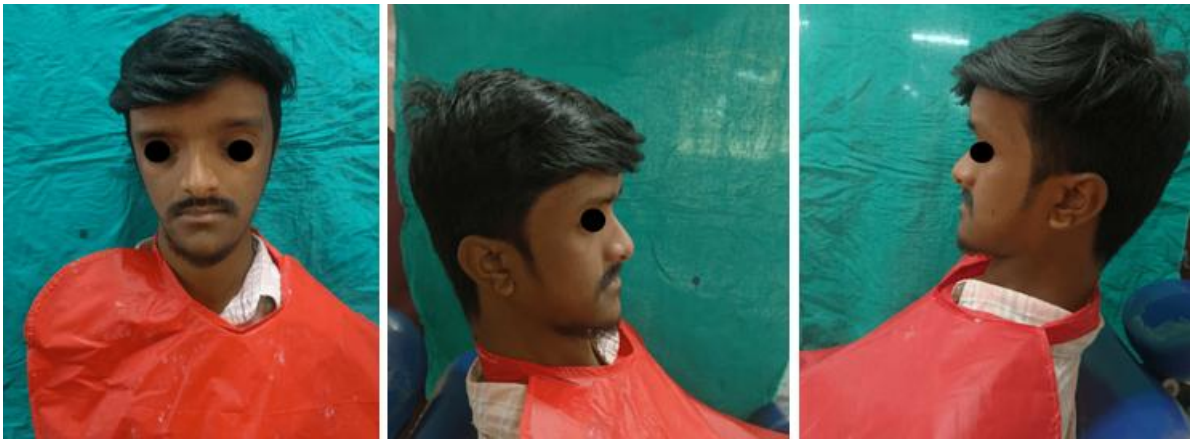


Figure 1: Flat face, occipital and parietal bossing, hypertelorism and sunken nasal bridge, hypoplastic mid-face

On intraoral examination decayed 84, 46. Multiple permanent were missing teeth with multiple retained deciduous teeth. Obliterated right and left buccal vestibule (Fig 2).

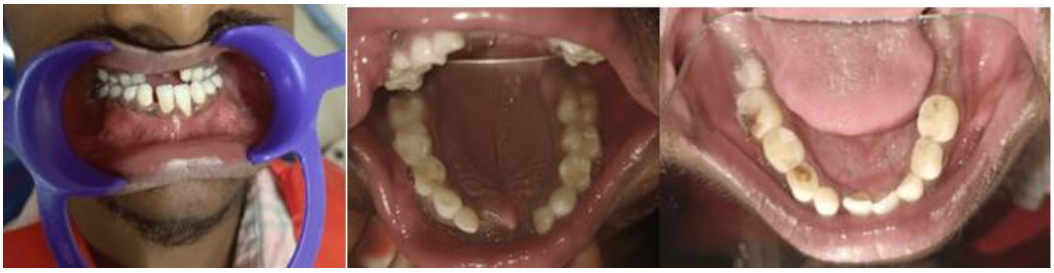


Figure 2: Intraoral view: showing multiple retained deciduous teeth, missing permanent teeth, and narrow, high arched palate, obliterated right and left lower buccal vestibule.

An oral pantamogram revealed multiple impacted permanent, supernumerary tooth, well defined peri-coronal radiolucency irt impacted 36, impacted supernumerary teeth (Fig 3).

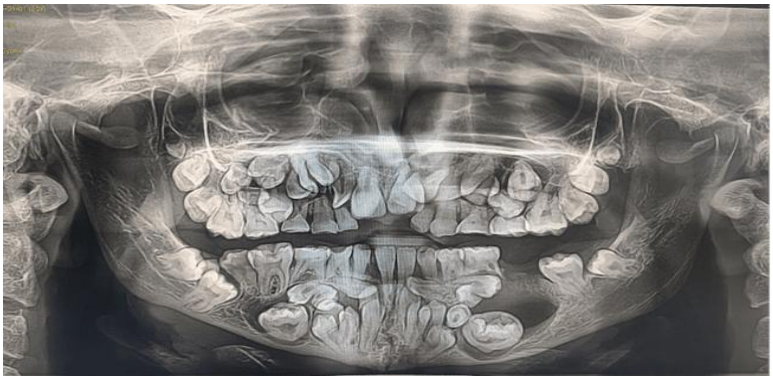
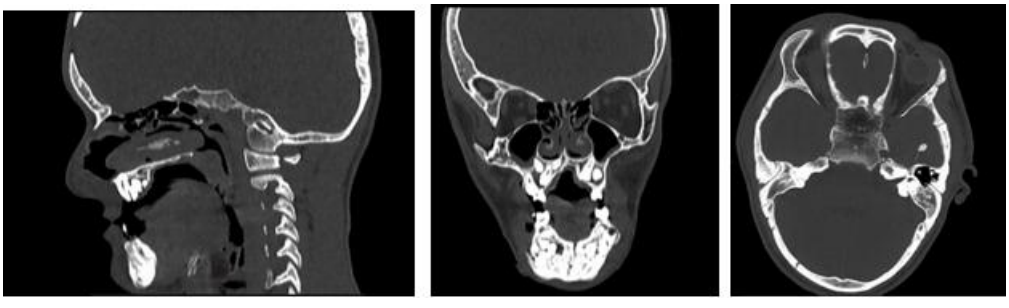


Figure 3: Orthopantomogram showing narrowing ascending ramus, slender pointed coronoid and multiple impacted teeth, cysts associated with it.

CT facial bones on coronal section, 3D sectioned CT images revealed open skull sutures; ossification of the zygomatic arch is incomplete, leading to discontinuous zygomatic arch (fig4). Both shoulders could not be approximated to midline either actively or passively, indicating an atypical presentation with normally developed clavicles (Fig. 5). Assessment of IQ was performed using the Binet Kulshreshtha test and suggested normal intelligence.



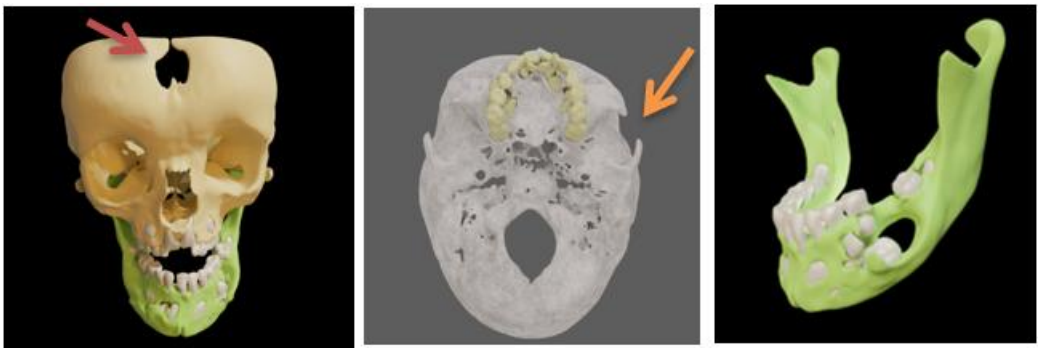


Figure 4: CT facial bones, 3D sectioned images showing open skull sutues, narrow ramus, and incomplete zygomatic arch.

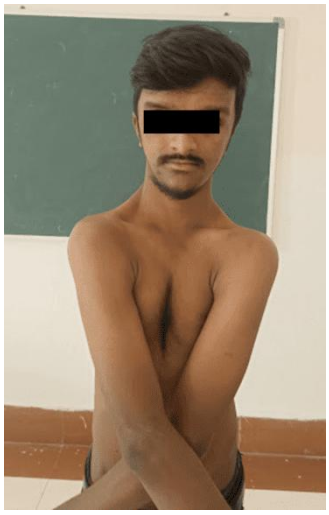


Figure 5: Both shoulders could not be approximated to midline, suggesting of normal clavicles

Chest radiographs demonstrated normally developed clavicles, ribs, and thoracic vertebral bodies. (Fig6) Radiographs of the hands depicted tapered distal phalanges of both thumbs, absence of tufting of distal phalanges of index fingers with second metacarpals showing accessory ossification centers at the bases (Fig7). The bone age was between 10 and 15 years as assessed by the Greulich and Pyle atlas.(7) Blood calcium, phosphate, alkaline phosphatase and thyroid profiles were normal. Sonographic examination of the abdomen and renal region revealed no abnormal findings.

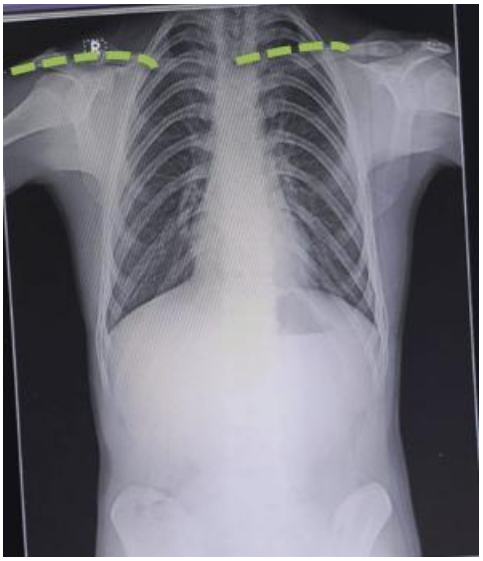


Figure 6: Chest radiographs demonstrated normally developed clavicles, ribs.

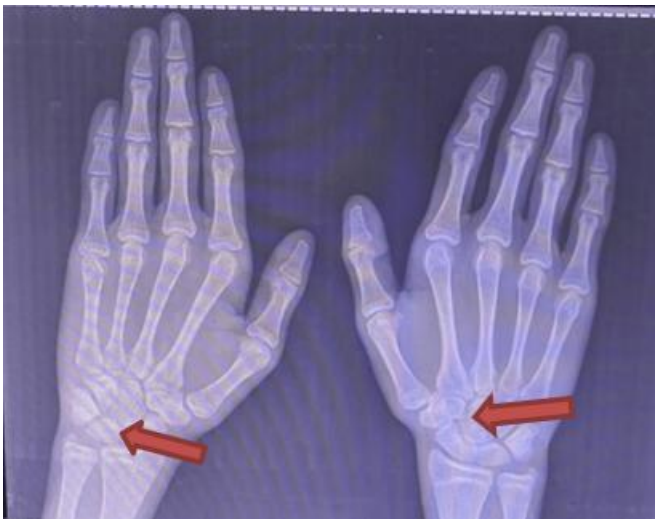


Figure 7: Psuedoepiphysis at the base of the second metacarpal

Cleidocranial dysplasia was diagnosed based on the typical clinical and radiographic features. Extraction of supernumerary teeth along with the cyst enucleation was performed. (Fig 8)

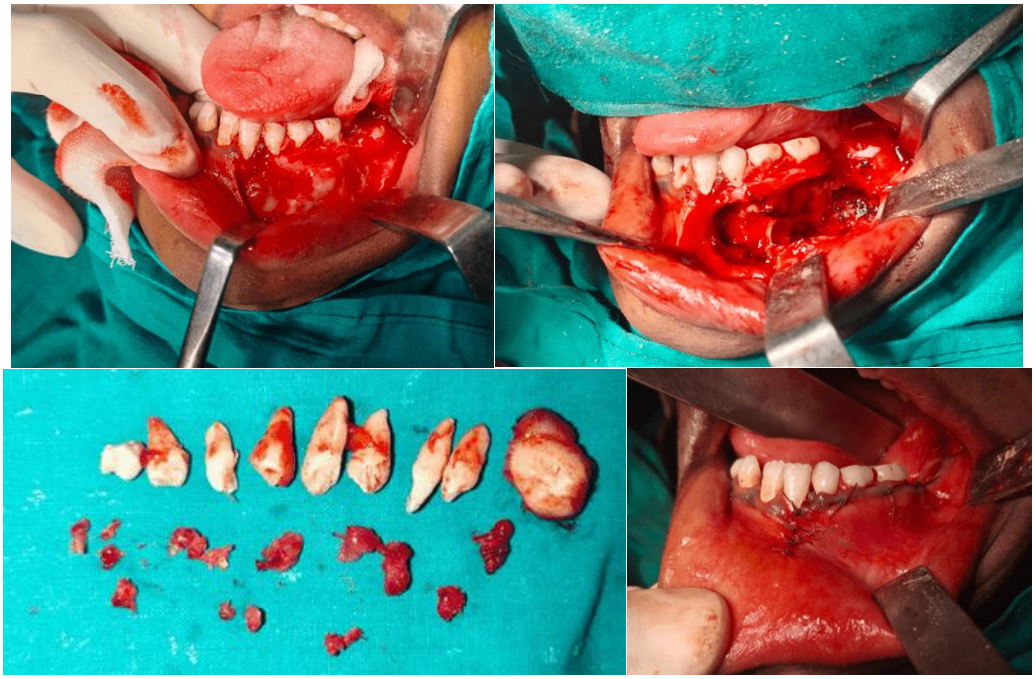


Figure 8: Enucleation of cyst, extraction of impacted supernumerary teeth

3. Discussion

Intermembranous and enchondral ossification bones are affected by CCD, a widespread skeletal abnormality.(8) Marie-Sainton disease, cleidocranial dysostosis, and mutational dysostosis are other names for CCD. Chromosome 6p2 has been identified as the gene responsible for this condition.(9) It is uncertain what causes cleidocranial dysplasia. There have been descriptions of both recessive and dominant inheritance patterns. Twenty to forty percent of the core binding factor α 1 (CDFA-1) gene on chromosome 6p21 undergoes spontaneous mutation.(10)

CCD involves the bones that are ossified earliest in fetal life; the clavicle is the first bone to show ossification. The disease classically causes a retardation or partial aplasia of bones that ossify intramembraneously (11) It is typified by anomalies of the jaws, clavicles, and skull in addition to sporadic long bone stunting. The patient is often short in stature, but the clavicle is either completely or partially aplastic on one or both sides, causing the neck to seem comparatively long and the shoulders to droop. It is more common to have partial aplasia.

There were also several wormian bones, open fontanelles and skull sutures, pelvic abnormalities, delayed closure of the pubic symphysis, and congenital hip dislocation.(12) (13) here we reported patient of 21 years with open skull sutures, normal clavicles. The vast variation in the condition's expression, which is controlled by the particular kind and position of the RUNX2 gene mutation, is the main cause of normal clavicles.

The dental abnormalities include several supernumerary teeth and the failure of the permanent dentition to erupt. Both jaws contain supernumerary teeth, particularly in the premolar regions.(14) The anterio-posterior and vertical dimensions of the central portion of the face is hypoplastic.(15) The hard palate is often narrow and high. Ectopic eruption of permanent teeth and the development of cysts connected to impacted teeth are further significant dental findings. (16) Unerupted teeth in CCD are thought to be caused by either (i) a disruption of bone resorption, (ii) a lack of cellular cementum, or (iii) a lack of union between the dental follicle and the mucosa because of intervening fibrous tissue that acts as an eruption barrier.

The most crucial tool for confirming the confirmation of CCD is the radiograph. A patient with CCD has an aplastic or hypoplastic clavicle on their chest radiograph. Skull radiographs are most crucial to diagnose this condition. Diffuse regions of rarefaction can be seen in the skull, with the frontal bones having the most ossification and the temporal and parietal bones having the least. The fontanelles are oversized and continue into maturity, and the cranial sutures are broad with abnormal suture lines. (17)The paranasal sinuses are often small and undeveloped. Mastoid air cells may not pneumatize at all. The skull bones of affected individuals often exhibit supplementary centres of ossification, giving the impression that there are a lot of wormian bones in between the main cranial bones. In our present case report, the same characteristics were noted. Radiographically, there are numerous impacted teeth in the both jaws which are significantly out of alignment. The high incidence of supernumerary teeth in maxillae and mandible occurs at premolar region.(17) In our case except 1st molars and mandibular central incisors remaining all the permanent teeth, along with supernumerary teeth are impacted and these impacted teeth are associated with dentigerous cysts.

Laboratory findings like thyroid function tests, haematological investigations are usually within normal limits.(17) In our case we have observed the same.

Correction of dentofacial abnormalities, both functionally and aesthetically is still being explored in the dental literature, despite the fact that there is no specialized treatment for patients with CCD. The patient's age has an impact on treatment plans. When an early diagnosis is made, the

supernumerary teeth are surgically removed, and brackets are used to apply orthodontic traction until the teeth achieve a suitable occlusion.(18)

A multidisciplinary approach must be considered in the treatment of CCD.(19) The time of the diagnosis of CCD may also be suggested as a guide for the choice of the necessary treatment model.

In our present case surgical management by enucleation of cysts followed by extraction of impacted teeth was done.

This case contributes to the limited literature on phenotypic variability in CCD, particularly highlighting that the presence of normal clavicles does not preclude diagnosis.

4. Conclusion

This case emphasizes the phenotypic diversity of cleidocranial dysplasia by showcasing an unusual appearance with normal clavicles. Despite being a defining characteristic, hypoplastic or missing clavicles do not rule out the diagnosis. Accurate diagnosis still requires careful clinical evaluation, radiographic assessment, and genetic investigation, especially in patients with craniofacial and dental symptoms associated with CCD. The identification of these alternative presentations expands our knowledge of the illness spectrum and emphasizes the significance of taking CCD into account even in the absence of traditional skeletal abnormalities.

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