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Case Report

Dentistry Section

Cleidocranial Dysplasia: A Case Report Illustrating Diagnostic Clinical and Radiological Findings

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ABSTRACT

Cleidocranial dysplasia (CCD) is a rare congenital defect, primarily affecting bones, undergoing intramembranous ossification. CCD presents with skeletal defects of several bones, the most striking feature of which are partial or complete absence of clavicles, late fontanels closure, and presence of open skull sutures and multiple wormian bones. These patients may be first noticed by the dentist because of the aesthetic problems or delayed eruption of teeth experienced by the patient. Here, we report a typical case of CCD in a 15-year-old female who had classical diagnostic feature of this syndrome.

Keywords: Bone defects, Cleidocranial dysplasia, Clavicular defect, Supernumerary teeth, Strawberry tongue

CASE REPORT

A 15-year-old female patient reported to the Department of Oral Medicine and Radiology for routine dental checkup. Her family history revealed that they were 1 brother and 2 sisters. There was history of consangious marriage. Patients mother also stated that the patient's mental status was not upto the mark and she was dull in grasping things.

The general physical examination revealed a moderately built and moderately nourished patient. On extraoral examination, the patient presented with flat face and occipital, parietal bossing, hypertelorism and sunken nasal bridge [Table/Fig-1]. Open fontanelle could be palpated in the anterior region of the head. Shoulders appeared narrow and showed marked drooping. On palpation, clavicles were absent bilaterally. Patient was able to approximate both shoulders in the midline on the chest [Table/Fig-2]. Intraoral examination revealed the presence of a set of deciduous dentition with all first permanent molars present and narrow high arched palate [Table/Fig-3]. A strawberry colored, fissuring on the dorsal anterior 2/3rd of tongue was observed [Table/Fig-4].

On orthopantomogram mandible demonstrated coarse trabeculation with areas of increased density, narrow ascending ramus and slender pointed coronoid processes and impacted central incisors, lateral incisors, canines, first & second premolars [Table/Fig-5]. Posteroanterior skull showed open frontanellae and open suture bones with presence of many wormian bones [Table/Fig-6].

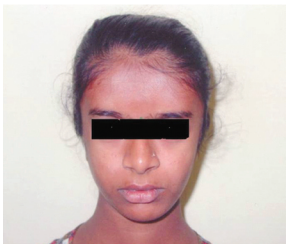
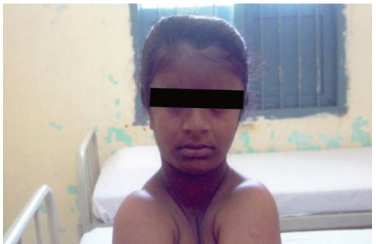
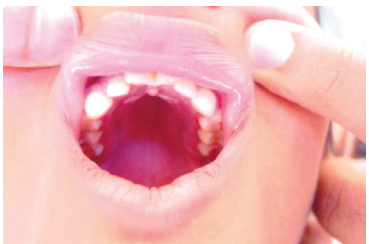
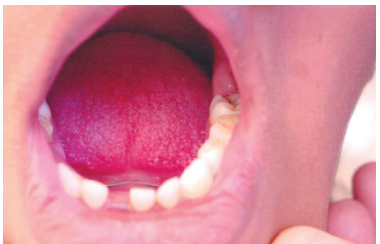
Paranasal sinus view showed that sinuses were poorly developed and only lateral sinus could be made out [Table/Fig-7]. Lateral cephelogram revealed a hypoplastic maxilla, with multiple impacted teeth and incomplete development of mastoid air cells [Table/Fig-8]. The posteroanterior view of chest radiograph showed complete bilateral absence of clavicle [Table/Fig-9]. Histopathological ground section revealed total lack of cellular cementum [Table/Fig-10,11].

Based on the typical clinical examination and radiographic feature, a diagnosis of cleidocranial dysplasia was made.

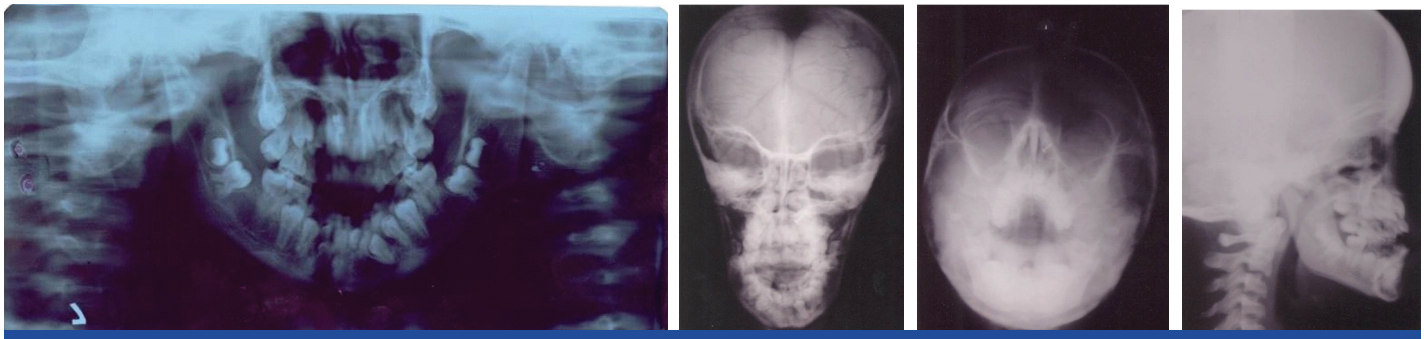
DISCUSSION

Cleiodocranial dysplasia (CCD) which is generally accepted as an autosomal disorder, was first described in 1765 by Martin [1], and was termed 'cleidocranial dysostosis' in 1897 by Marie and Sainton [2]. CCD is also known as Marie-Sainton disease, mutational dysostosis, and cleidocranialdysostosis. A gene for this disorder has been mapped to chromosome 6p21 [3,4]. The aetiology of cleidocranial dysplasia is unknown. Both dominant and recessive patterns of inheritance have been described. Spontaneous mutation occurs in 20-40% cases in the core binding factor α 1 (CDFA-1) gene, located on chromosome 6p21 [5]. It may be discovered at any age, but the cranial deficiencies may be noticed at birth. Both sexes are affected to an approximately equal extent. The defect often appears in several successive generations [6].

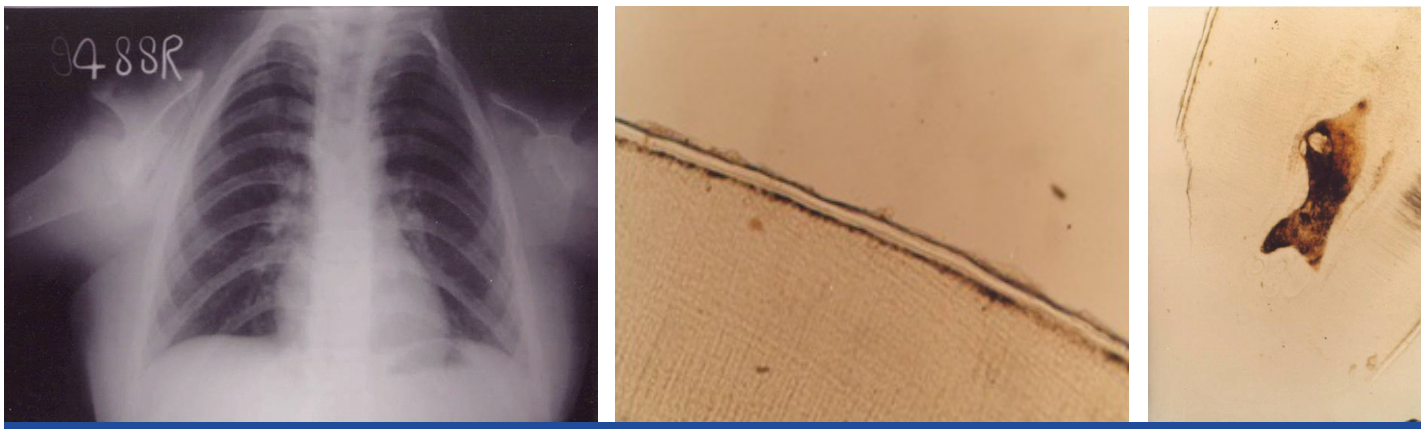
CCD involves the bones that are ossified earliest in fetal life, the clavicle is the first bone to show ossification. The disease classically causes a retardation or partial aplasia of bones that ossify intramembraneously [4,7] Here, we report a case of 15-year-old female manifesting the classical features of CCD. (CCD) is a relatively uncommon disorder with a prevalence of 0.5 per 100,000 live births [8]. It is characterised by abnormalities of the clavicles, skull and jaws as well as by occasional stunting of long bones. The stature of the patient is usually short, but the neck appears relatively long and the shoulder droop results from the clavicle being unilaterally or bilaterally totally or partially aplastic. Partial aplasia is more common

[Table/Fig-1]: Showing flat face, occipital and parietal bossing, hypertelorism and sunken nasal bridge
[Table/Fig-2]: Showing approximation of shoulder
[Table/Fig-3]: Intraoral view: showing a narrow, high arched palate
[Table/Fig-4]: Intra-oral view showing strawberry colored tongue



[Table/Fig-5]: Orthopantomogram narrow showing ascending ramus,slender pointed coronoid and impacted teeth
[Table/Fig-6]: Posteroanterior skull view revealing open frontanellae and open suture bones with presence of many wormian bones
[Table/Fig-7]: Paranasal sinus view showing poorly developed sinuses
[Table/Fig-8]: Lateral cephalogram showing hypoplastic maxilla, with multiple impacted teeth and incomplete development of mastoid air cells



[Table/Fig-9]: PA view chest showing complete bilateral absence of clavicle
[Table/Fig-10]: Histopathological ground section revealing lack of cellular cementum
[Table/Fig-11]: Histopathological ground section showing lack of cellular cementum

at the acromion ends. Congenital dislocation of hip, delayed closure of pubic symphysis, pelvic dysplasia, open fontanelles and skull suture and numerous wormian bones were also seen [9,10]. The same features were reported in our case.

The oral manifestations are failure of eruption of the secondary dentition, and multiple supernumerary teeth [11]. Supernumerary teeth are localized in both jaws especially in areas of premolars [12]. The middle third of face is hypoplastic both in the anterior posterior and vertical dimensions. Hard palate is generally high and narrow. Other intraoral manifestations are ectopic eruption of permanent teeth and the formation of cyst associated with impacted teeth [13]. The cause of unerupted teeth in CCD is said to be (i) a disturbance of bone resorption (ii) a lack of cellular cementum or (iii) a lack of union between the dental follicle and the mucosa due to interposed fibrous tissue acting as a barrier to eruption.

The radiograph is the most important means by which the diagnosis of CCD can be confirmed [12]. Chest radiograph of patient with CCD shows hypoplastic or aplastic clavicle. Radiographs of skull are pathognomic of the disease. The skull shows diffuse areas of rarefaction with most ossification in the frontal bones and the least in the temporal and parietal bone. Cranial sutures are broad with anomalous suture lines and the fontannels are large and persists into adulthood. Paranasal sinuses are usually underdeveloped and narrow. Pneumatization of mastoid air cells may not occur at all. The individual bones of the head frequently shows evidence of accessory centers of ossification which gives the appearance of large numbers of wormian bones present between the main cranial bones. The same features were reported in our case. Radiographically, the mandible and maxilla contain many unerupted teeth that are greatly deviated from the normal position. Supernumerary teeth are often present and most frequently seen in the premolar site [12].

Laboratory findings are usually within normal limits. The calcium' and phosphatase levels of the blood are normal but the phosphorus level may be low. In the majority of cases, results of blood analysis have been within normal limits [12].

Grade 3 mobility was present in relation to 81, therefore, we planned for histopathological tooth section and they reported a lack of cellular cementum. The same finding was reported by Yamamato H et al., [11], and they also suggested it as a possible cause for delayed eruption of teeth.

Although, there is no specific treatment for the patient with CCD, the correction of the dentofacial deformities both aesthetically and functionally is still being discussed in the dental literature. Treatment planning is related to the age of the patient. In cases which are diagnosed at early ages, the supernumerary teeth are surgically exposed and orthodontic traction is applied by the help of brackets until the teeth come into acceptable occlusion [13]. Trimble et al., [14] suggested orthognathic surgery for the correction of middle - third of the face. Winter has advocated the extraction of all the primary and permanent teeth and placement with a removable prosthesis [13]. The main problem with this treatment is alveolar hypoplasia which compromises of the retention of prosthesis. Pusey & Durie [15] suggested removal of only the erupted teeth and use of a removable prosthesis to minimize the alveolar bone loss. It is not always easy to determine the most appropriate aesthetic and functional treatment model for dentofacial deformities in patient with CCD. A multidisciplinary operation must be considered in the treatment of CCD and a long time approach must be planned [16]. The time of the diagnosis of CCD may also be suggested as a guide for the choice of the necessary treatment model.

REFERENCES

[1] Martin M. Sur undeplacement natural de la clavicule. *J Med ChirPharmacol*. 1765; 23 :456.
[2] Marie P, Sainton P. Observation d'hydrocephalic hereditary (pereetfils), par vice de development du crane et du cerveau. *Bull Soc Med Hop Paris*.1897;14:706.

[3] Mundlos S, Mulliken JB, Abramson DI, Warman ML, Knoll JHM & Olsen BR. Genetic mapping of cleidocranial dysplasia and evidence of a microdeletion in one family. *Hum Mol Genet*.1995;4:71-75.
[4] Mundlos S. Cleidocranial dysplasia: clinical and molecular genetics. *J Med Genet*. 1999; 36(3):177-82.
[5] Fitchet SM. Cleidocranial Dysostosis: hereditary and familial. *J Bone Surg.* 1929; 11: 838 -66.
[6] Reddy SK, Parmar R. Cleidocranial Dysplasia (Dysostosis):A Case Report. *The Orthodontic CYBER Journal*. 2010 March. Available from: orthocj.com/2010/03/cleidocranial-dysplasis-dysostosis-a-case-report/.
[7] Currall V, Clancy R, Diamond D. Cleidocranial dysplasia. *Curr Orthop*. 2007; 21(2): 159-62.
[8] Yochum TR and Rowe LJ. Essentials of skeletal Radiology, Williams & Wilkins, Baltimore, Md, USA, 2nd Edition, 1996.
[9] Mohan RP, Suma GN, Vashishth S, Goel S. Cleidocranial dysplasia: Clinico-radiological illustration of a rare case. *J Oral Sci*. 2010; 52(1): 161-66.
[10] Tanaka JL, Ono E, Filho EM, Castilho JC, Moraes LC, Moraes ME. Cleidocranial dysplasia: importance of radiographic images in diagnosis of the condition. *J Oral Sci*. 2006; 48(3): 161-66.
[11] Yamamoto H, Sakae T, Davies JE. Cleidocranial Dysplasia: A light microscope electron microscope and crystallographic study. *Oral Surg Oral Med Oral Path*. 1989; 68: 195 - 200.
[12] Koch PE, Hammer WB. Cleidocranialdysostosis: review of literature and report of case,*J Oral Surg*. 1978; 36: 39-42.
[13] FerdaTasar, EnginBelut, Celal Turner, Mustafa Saysel, MehinetMuhtarogullan. Clediocranial dysplasia: case report. *Austr Dent J*. 1995; 40(6): 352-56.
[14] Trimble LD, West RA, McNeill RW. Cleidocranialdysplasia. Comprehensive treatment of thedentofacial abnormalities. *Jam Dent Assoc*. 1982; 105: 661-6.
[15] Pusey RF, Durie JF. A case of cleidocranialdysostosis showing failure of eruption of teeth. *Br Dent J*. 1943; 75(1): 11-13.
[16] Ozkan G, Kanli A. Clinical and radiological manifestations of cleidocranial dysplasia: A Case report. *Indian Journal of dentistry*. 2013; 4(3):174 -77.

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