International Journal of Applied Dental Sciences 2019; 5(3): 646-649

ISSN Print: 2394-7489
ISSN Online: 2394-7497
IJADS 2020; 5(3): 646-649

www.oraljournal.com
Received: 08-02-2020
Accepted: 10-03-2020

Prenika Sharma
Postgraduate Student, Department of Oral Medicine and Radiology, Govt. Dental College, Srinagar, Jammu and Kashmir, India

Altaf Hussain Chalkoo
Professor and HOD, Department of Oral Medicine and Radiology, Govt. Dental College, Srinagar, Jammu and Kashmir, India

Correspondence
Prenika Sharma
Postgraduate Student, Department of Oral Medicine and Radiology, Govt. Dental College, Srinagar, Jammu and Kashmir, India

Cleidocranial dysplasia- Clinico-radiological presentation of 2 cases

Prenika Sharma and Altaf Hussain Chalkoo

Abstract
Cleidocranial dysplasia or cleidocranial dystosis is an autosomal dominant syndrome, best known for its dental and skeletal abnormalities. The term cleido refers to clavicle; cranial refers to head; and dysplasia means "ill formed" (Feldman, 2002). It is caused by mutations in major regulator of bone differentiation which is known as transcription factor RUNX2. In first case a 18year old female patient visited the department as case of missing upper front teeth. On clinical examination features were suggestive of cleidocranial dysplasia which was later confirmed with radiological investigations for rudimentary clavicles and impacted supernumerary teeth and open fontanelles. There was no such family history present. In the second case patient had similar history in father and paternal uncle.

Keywords: Cleidocranial Dysplasia (CCD), supernumerary teeth, rudimentary clavicle, MAD (Mandibuloacral dysplasia)

1. Introduction
Cleidocranial dysplasia (CCD) is a well-defined skeletal disorder with characteristic clinical findings. Martin in 1765 reported it for the first time. It shows autosomal dominant inheritance, it affects 1 in 1,000,000 live births (Jones, 1997) by Woshnik et all. According to clinical manifestations, CCD can be classified as typical CCD, light CCD, and isolated tooth dysplasia [1].
Individuals with CCD are typically of short stature, with brachycephalic skull and bossing of the frontal and parietal bones. Hypoplasia of the mid-face and mandibular prognathism is present. The sutures and fontanelles of the skull exhibit delayed closure with the occurrence of secondary centers of ossification in these areas and the formation of Wormian bones [2]. here we presented clinical and radiological features of typical CCD cases, adding to the literature.

Case 1:
Presented case is 18 year old female patient. She complains of delayed eruption of missing teeth in upper front region of jaw.
Detailed medical history, clinical and radiological examination, revealed her a case of cleidocranial dysplasia. No other family member in her family is affected.
Patient is of short stature from her age, thin and lean in built. Patient was able to approximate her shoulders in the center (figure 1). Frontal and parietal bossing is present with open anterior fontanelles and open metopic and sagittal sutures (figure 2). Brachycephalic shape of the head present with normal hairs. Nasal bridge is depressed and hypertelorism is present. Maxillary hypoplasia and mandibular prognathism giving rise to concave profile. Short tapered fingers and broad thumbs (figure 5).
On Intraoral examination, missing left maxillary anterior teeth, narrow, high arched palate, root stumps of left maxillary and mandibular first molar tooth. Crown of Supernumerary tooth present erupting lingually in relation to mandibular right canine. (Figure 3,4)
Radiological investigation included intraoral periapical radiograph and orthopantomogram (OPG), it showed impacted 4 permanent and 4 supernumerary teeth in anterior maxillary region. Maxillary sinuses are underdeveloped. Discontinuous clavicles, narrow thorax, oblique ribs present on chest X-ray. (Figure 6)
On AP and Lateral skull view present open anterior fontanelles, persistent part of metopic and

sagittal sutures, prognathic mandible present.(figure 7, 8)

Fig 1: Patient's stature was short compare to her age.

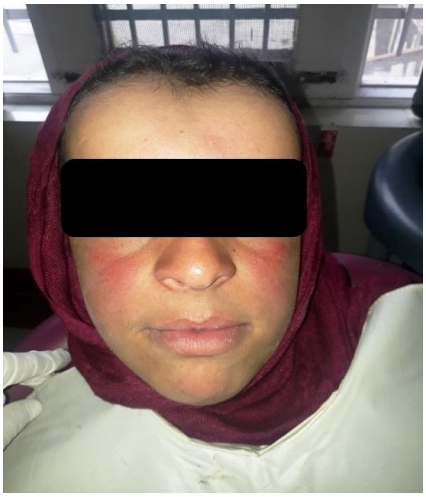

Fig 2: Depression is present due to open anterior fontanelles. Nasal bridge is depressed with hypertelorism.

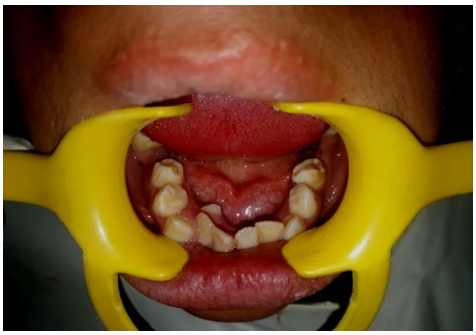

Fig 3: Mandibular crowding present

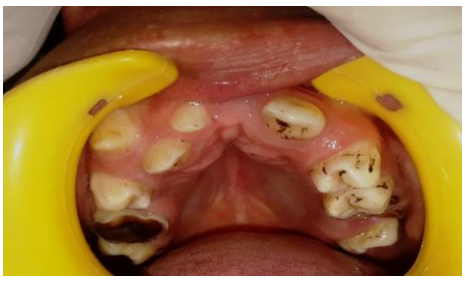

Fig 4: High arch palate with crowding in maxillary teeth.

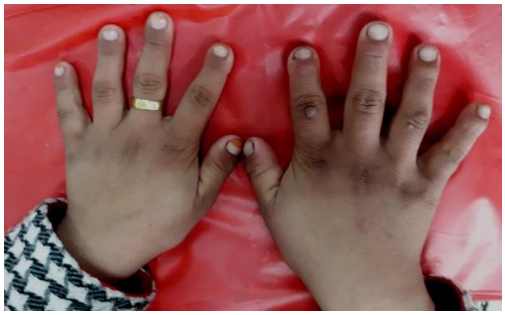

Fig 5: Short tapered fingers and broad thumbs.

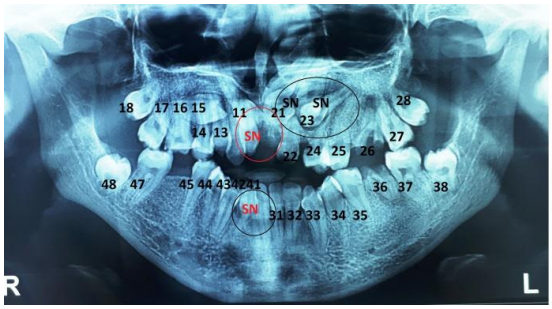

Fig 6: Opg showing impacted 4 permanent and 4 supernumerary teeth in anterior maxillary region. Maxillary sinuses are underdeveloped

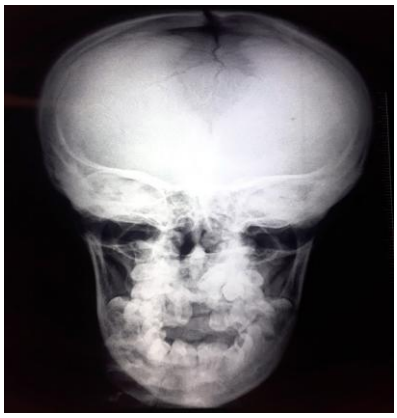

Fig 7: On AP skull view presents open anterior fontanelles, persistent part of metopic

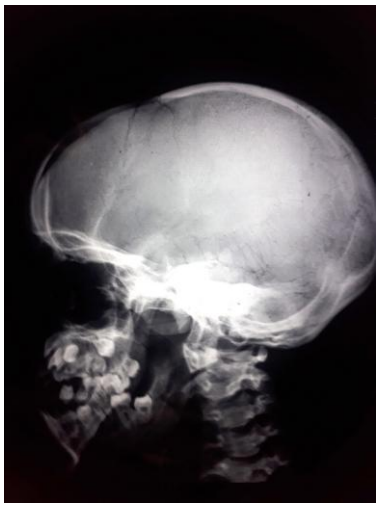

Fig 8: On lateral skull view presents Wormian lines and prognathic mandible.

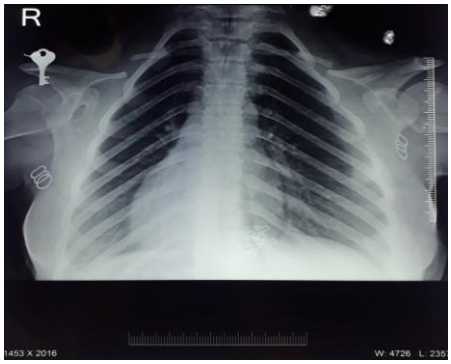

Fig 9: X Ray chest shows discontinuity in the clavicles on both sides.

Case 2:

Presented case is a 13yr old male patient non-diabetic, non - hypertensive visited our department with a chief complaint of missing teeth in the mouth. He was in his usual state of health, his father told that his milk teeth did not shed on time in upper front region and missing teeth in right and left lower back region of jaw. They consulted dentist in sub- district hospital, who then referred him here for further management. There is family history of same problem in father and paternal uncle. On examination patient was able to approximate his shoulders in the center. Frontal and parietal bossing is present with open anterior fontanelles. (Fig 10) Brachycephalic shape of the head present with normal hairs. Nasal bridge is depressed and hypertelorism is present. Concave profile, maxillary hypoplasia and mandibular prognathism. Short tapered fingers and broad thumbs present. A-P View skull showing open anterior fontanelles and Wormian bones. OPG showing multiple retained permanent teeth and supernumerary teeth (Fig 11, 12). X- Ray chest showing hypoplastic clavicles both sides. (Fig 13)

Intraoral examination showed multiple missing teeth and high arch palate. (Fig 14)

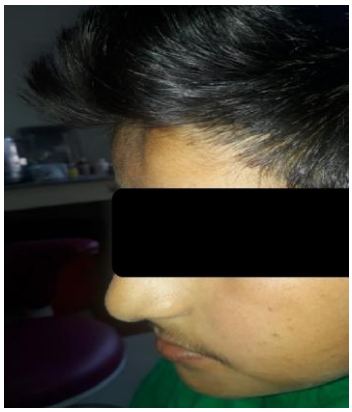

Fig 10: Showing depression in the place of anterior fontanelle.

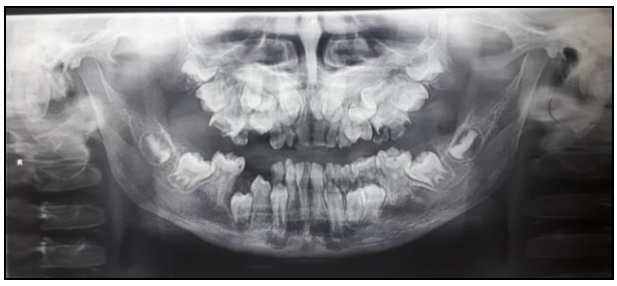

Fig 11: OPG showing multiple retained permanent and supernumerary teeth**.**

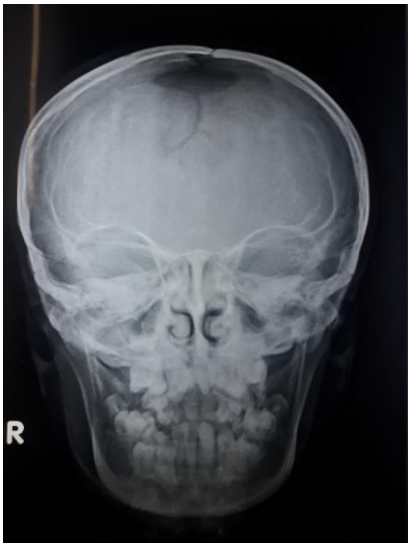

Fig 12: Showing AP Skull view with open anterior fontanelle and Wormian bones

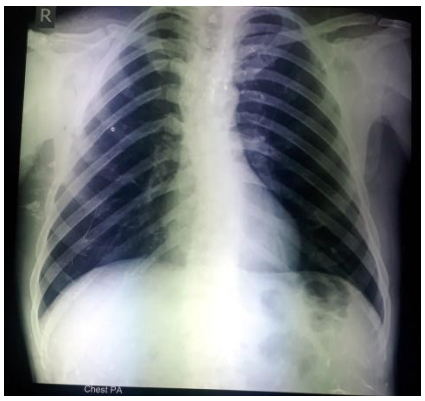

Fig 13: X Ray Chest showing discontinuity in the clavicles

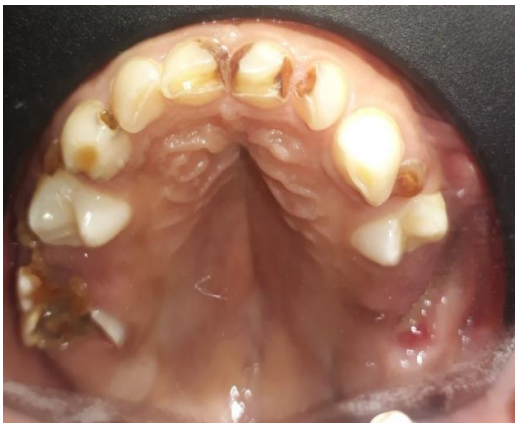

Fig 14: Intraoral picture showing retained deciduous teeth.

2. Discussion

Cleidocranial dystosis is also called as Marie and Sainton's disease, Mutational dysostosis. The locus for CCD has been mapped to chromosome 6p21 (Lee *et al*., 1997) [3]

It is seen that certain degrees of clavicular hypoplasia is a consistent feature of this disorder. There is involvement of the facial bones, prolonged retention of deciduous teeth, impacted permanent successors and supernumerary elements, occasionally follicular cysts and eruptive pseudo cysts also present [4].

The highest number of supernumerary teeth reported in a patient with cleidocranial dysplasia in literature is 63 and here we reported 4 supernumerary teeth [5]. Complete absence of

the clavicle permitting an abnormal mobility of shoulders seen in about 10% of cases.

Pyknodysostosis and cleidocranial dysplasia shares many features; can be differentiation by the absence of supernumerary teeth and increased bone density on the radiograph (Maroteaux and Lamy, 1962). Mandibuloacral dysplasia is an autosomal recessive disorder. It can be differentiated from CCD by progressive stiffening of joints and radiographs reveal acroosteodysplasia of the fingers and toes, with delayed ossification of the carpal bones (Novelli *et al*., 2002) [6].

Dental management of the patient depends on the chronological and dental age [7]. It requires multidisciplinary approach. Jensen and Kreiborg recommended early diagnosis of CCD to initiate the early treatment in order to minimize the extent of later surgical and orthodontic intervention. Dental treatment is difficult and time consuming, often requiring surgical, orthodontic and prosthodontics interventions.

Various treatment options are suggested for treatment such as removal of the impacted permanent, supernumerary and primary teeth followed by fabrication of over-dentures. Others suggested orthodontic traction of the impacted permanent teeth; removal of the supernumerary teeth immediately after completion of mineralization of their crowns [7, 8].

The present case we saw multiple missing permanent teeth in oral cavity and multiple impacted teeth on OPG, this highlights the role of dental physician in early diagnosis and treatment of CCD, to minimize psychological trauma to these patients in future. Patient should be kept on follow up to monitor any cystic changes in the impacted teeth.

3. References

1. Woshnik KK. Cleidocranial dysplasia: the lived experience (doctoral dissertation, brigham young university. college of nursing).
2. Zi-Jian Li, Jun-Yan Wang, Ming-Fei Gao, Da-Lei Wu And Xin Chang. Orthodontic treatment of a patient with cleidocranial dysplasia: A case report. Experimental and therapeutic medicine 2016; 12:690-694,
3. Wang GX, Sun RP, Song FL. a novel runx2 mutation (t420i) in chinese patients with cleidocranial dysplasia. genetics and molecular research. 2010; 9(1):41-47
4. Zi-Jian LI, Jun-Yan Wang, Ming-Fei Gao, Da-Lei Wu1, Xin Chang. orthodontic treatment of a patient with cleidocranial dysplasia: a case report. Experimental and therapeutic medicine. 2016; 12:690-694.
5. Yamamoto H, Sakae T, Davies JE. Cleidocranial dysplasia: A light microscope, electron microscope and crystallographic study. Oral Surg Oral Med Oral Pathol 1989; 68:195-200,
6. Pradhuman Verma, Kanika Gupta Verma, Som Datt Gupta. Cleidocranial dysplasia: a dilemma in diagnosis. Archives of Orofacial Sciences. 2010; 5(2):61-64
7. Kusum Bharti, Mridula Goswami. Cleidocranial dysplasia: A report of two cases with brief review. Intractable & Rare Diseases Research. 2016; 5(2):117-120.
8. Jensen BL, Kreiborg S. Development of the dentition in cieidocranial dysplasia, J Oral Pathol Med. 1990; 19:89-93.