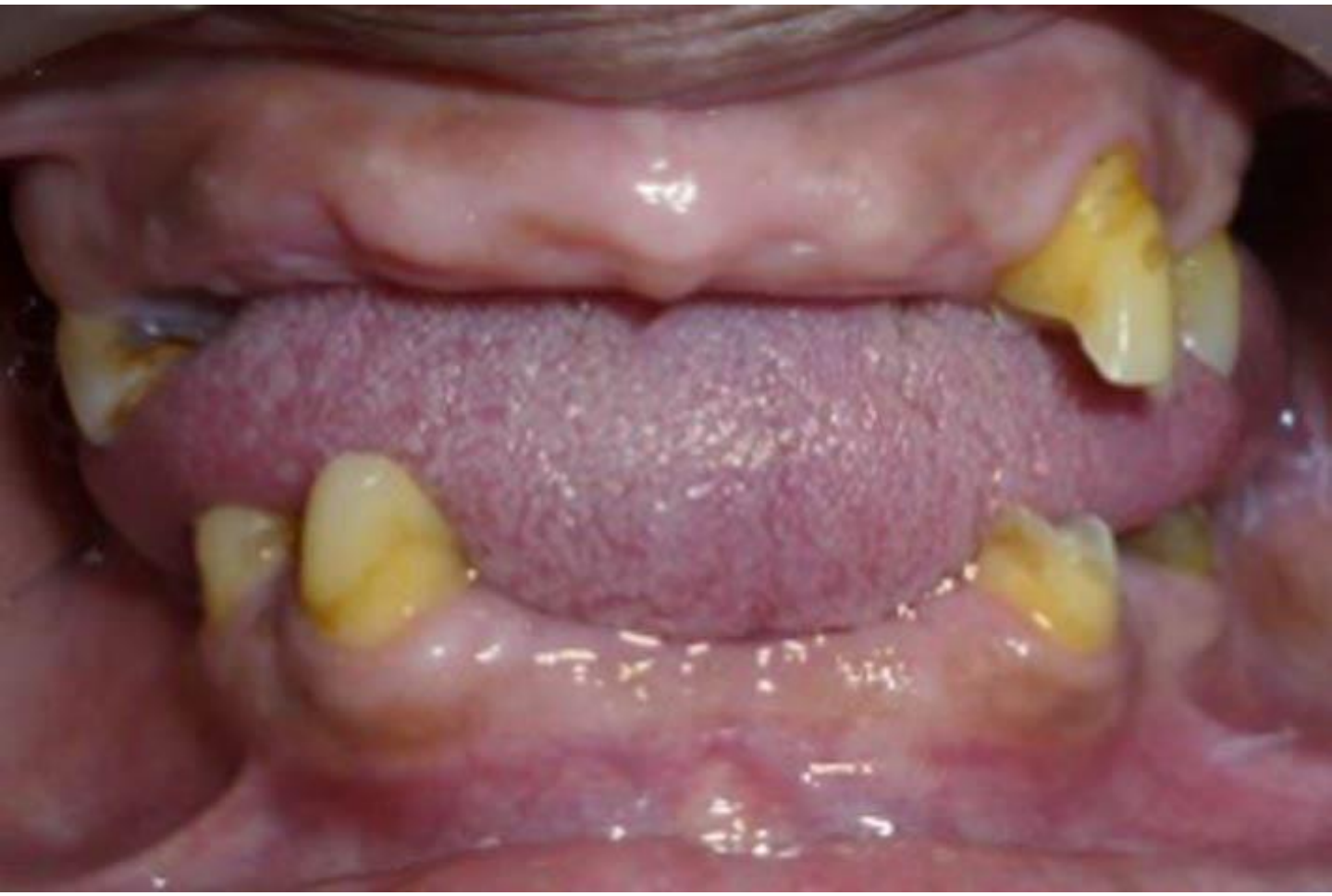
International Journal of Oral Care & Research

Volume 2 Issue 5 (Jul - Sep 2014) ISSN 2347-6249



An Official Publication of
"Ivano-Frankivsk National Medical University, Ukraine"
Officially Associated with
"The Egyptian Society of Oral Implantology"
"International Group for Oral Rehabilitation, France"

Case Report

Received : 03-05-14
Review completed : 28-06-14
Accepted : 06-07-14

CLEIDOCRANIAL DYSPLASIA: A CASE REPORT

Amol Dhokar, * Meghana SM, ** Monica Yadav, ***
Kaveri Chakravorty †

* Reader, Department of Oral Medicine & Radiology, Terna Dental College & Hospital, Nerul, Navi Mumbai, India
** Reader, Department of Oral Pathology & Microbiology, Terna Dental College & Hospital, Nerul, Navi Mumbai, India
*** Professor & Head, Department of Oral Pathology &Microbiology, Terna Dental College & Hospital, Nerul, Navi Mumbai, India
† Intern, Terna Dental College & Hospital, Nerul, Navi Mumbai, India

ABSTRACT

Cleidocranial dysplasia (CCD) is a rare autosomal dominant disease with a wide range of clinical variability. Dentists are often the first to encounter such patients, some of whom may not even show typical manifestations. CCD is caused by mutation in the gene located on 6p21, encoding transcription factor CBFA1, i.e. runt-related transcription factor 2 (RUNX2). The most prevalent features associated with CCD are aplastic or hypoplastic clavicles, supernumerary teeth, failed eruption of permanent teeth, and hypoplastic maxilla. We report a case of CCD in a 13 year old girl who was referred to our hospital for correction of mandibular prognathism and spacing between teeth. Clinical examination revealed classic features of CCD which were further confirmed after extensive radiographic survey. The details of this case are herein reported because of the extremely low incidence of this disorder.

KEYWORDS: Cleidocranial dysplasia; mutation; aplastic clavicles; retained deciduous teeth

INTRODUCTION

Cleidocranial dysplasia (CCD) is a rare disorder with autosomal dominant inheritance, though 40% cases occur spontaneously with no apparent genetic cause.[1] The incidence of the disorder has been reported to be 1:1000,000.[2] This disorder primarily affects bones which undergo intramembranous ossification. It is characterized by delayed closure of fontanelles, presence of open skull sutures, hypoplastic or aplastic clavicles, supernumerary teeth, delayed eruption of permanent dentition, wide pubic symphysis, short stature and a variety of other skeletal changes. A prominent feature of CCD is that the clavicular aplasia or agenesis allows approximation of the shoulders in front of the chest. Other prominent skeletal features include: delayed ossification of skull, interposition of wormian bones in open fontanels that persist until adulthood. With regards to extremities, the most marked changes are located in hands. Distal phalanges are hypoplastic and pointed.[3] First described by Marie and Sainton in 1898, CCD, also known as Marie and Sainton disease, cleidocranial dysostosis or mutational dysostosis,[4] is associated with a spontaneous mutation in the gene which is essential for osteoblast and odontoblast differentiation as well as bone and tooth formation. The gene has been mapped to chromosome 6p218 within a region containing core-binding factor subunit alpha-1 (CBFA1), a member of the runt family of transcription factors Runt-related transcription factor 2 (RUNX2).[5] CCD is of clinical significance in dentistry due to the involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth. The lacrimal and zygomatic bones are reported to be underdeveloped.[5] There is prolonged retention of the deciduous teeth and subsequent delay in eruption of the succedaneous teeth. Sometimes this delay is permanent. The roots of the teeth are often short and thinner than usual and may be deformed. Other oral manifestations include: high, narrow, arced palate; underdeveloped maxilla and paranasal sinuses. Here, we report a case of CCD in a 13 year old girl who desired orthodontic treatment and presented with the classical symptoms of the syndrome.

CASE REPORT

A 13 year old girl was referred to the institution with a chief complaint of prognathic mandible and desired correction of the same. Her parents reported no relevant medical history.

Fig. 1: Profile picture with excessive shoulder mobility. Also note brachycephaly, frontal bossing, hypertelorism, depressed nasal bridge and flat alar base

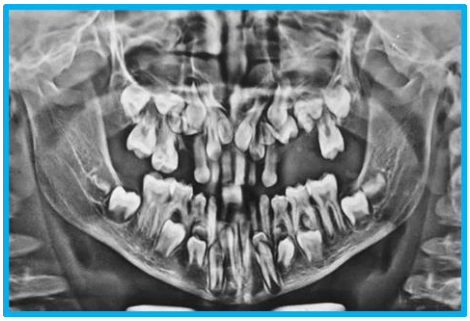


Fig. 2: OPG radiograph showing over retained primary and impacted permanent teeth

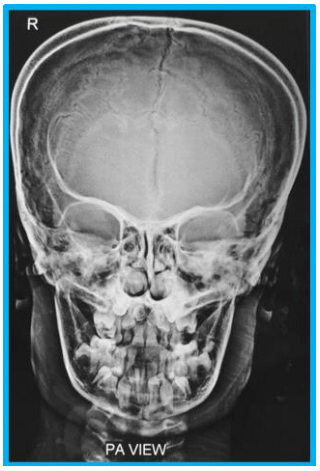


Fig. 3: PA skull radiograph showing open frontal suture, hypoplastic maxilla and underdeveloped maxillary sinuses

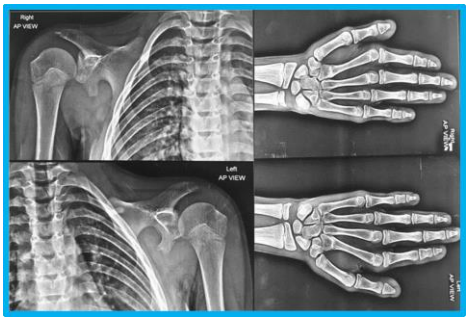


Fig. 4: Shoulder radiograph showing hypoplastic clavicles (left) and hand-wrist radiograph showing abnormally small last digits (right)

Her past dental history revealed extraction of the primary teeth 54, 64 and 65 two years ago due to caries and pain. Also, over-retained 71 and 81 were also extracted. On physical examination, the patient had disproportionate short stature with height measuring about 105 cms. She was well oriented with narrow and dropping shoulders. She exhibited hypertelorism, brachycephalic head with frontal bossing. Maxilla was hypoplastic and the nasal bridge was depressed with flat alar base. Further examination showed an abnormal facility in opposing her shoulders due to malformed or absent clavicle (Fig. 1) and a short fifth finger of both the hands. Intraoral examination revealed the persistence of deciduous dentition along with fully erupted permanent first molars. The palate appeared normal. She had class 3 skeletal profile and malocclusion. The panoramic radiograph confirmed the above intraoral findings. All the permanent teeth were present in the jaws but were impacted except for 16, 26, 36 and 46 (Fig. 2). PA skull radiograph showed open frontal sutures and smaller maxillary growth. The lateral cephalogram showed prognathic mandible, hypoplastic maxilla (Fig. 3) and skeletal class 3 growth. PA Water's projection revealed hypoplastic maxilla and maxillary sinuses. The PA chest radiograph showed hypoplastic clavicles (Fig. 4) and a narrow, bell-shaped thorax. The hand-wrist radiograph indicated short distal phalanges of the last finger. Based on these findings, a diagnosis of Cleidocranial Dysplasia was made.

DISCUSSION

CCD is an autosomal dominant malformation syndrome affecting bone and teeth. It affects both sexes equally. Until 2009, only 700 cases had been described since its original description.[6] It can be inherited or arise as a result of sporadic mutation. In approximately 40% of the CCD patients, a genetic transition code cannot be identified and this condition develops spontaneously.[7] The pathology relating to this condition is due to an early developmental disorder of mesenchyme or connective tissue. This causes retarded ossification of bone precursors, especially at junctions, which can lead to defective ossification, or even failure of ossification, of portions of the skeletal structure.[4] The dental abnormalities are often the reasons for diagnosis in mildly affected individuals. Dental

abnormalities are typical main features of CCD, and they occur in 93.5% of affected patients.[8] Characteristic patients with this disease show prolonged retention of the primary dentition and delayed eruption of permanent dentition. Extraction of primary teeth does not adequately stimulate eruption of underlying permanent teeth. There is paucity or complete absence of cellular cementum on both erupted and unerupted teeth. Often unerupted supernumerary teeth are present and considerable crowding and disorganization of the developing permanent dentition may occur.[9] Radiographic features of the jaws in an individual with CCD include smaller maxilla due to underdeveloped maxillary sinuses. The mandible is usually normal in size. There can be patent mandibular symphysis in some cases. The alveolar bone overlying unerupted teeth is denser than usual with coarse trabecular pattern in the mandible. This may account for delayed eruption of permanent teeth. Narrow high arched palate is found in some cases.[8] The unerupted supernumerary teeth develop in anterior and premolar region and resemble premolars. These unerupted teeth may develop dentigerous cysts later in some cases.[8] The diagnosis of CCD can be done with family history, hypermobility of shoulders, clinical examination of skull and pathognomic prolonged retention of primary teeth and multiple unerupted supernumerary teeth. However other conditions associated with multiple unerupted supernumerary teeth such as Gardener's syndrome and Pyknodystosis should also be kept in mind as differential diagnosis.[9] Absolute verification of diagnosis of CCD can only be obtained by molecular genetic analysis.[5] Even with some potential complications, the life span of an individual with CCD is normal.[3] There is no specific treatment of CCD, although care of other oral conditions is important. The over retained teeth should be restored if they become carious, since their extraction does not necessarily induce eruption of permanent teeth. However in recent years, researchers have suggested that removal of retained primary and supernumerary teeth may facilitate the eruption of permanent teeth. The bone overlying the normal tooth should be removed to expose the crown when half of the permanent tooth is formed. This will aid in eruption of permanent teeth.[9] Ideally patients should be identified early before 5 years of age, to take the advantage of orthodontic and surgical treatment.[9] Surgical exposure and orthodontic positioning can help to expand the maxillary arch to gain additional space for tooth alignment. Prosthodontic rehabilitation with implants has been used in some cases. Patients should be monitored for development of distal molars and cysts until adolscence.[8] Orthognathic surgeries and orthodontic alignment can help to improve the aesthetics.[10]

CONCLUSION

In conclusion, CCD although present since birth, could be easily missed because of its extremely low frequency and lack of typical syndromic manifestations in early childhood. Hence the dentist plays a vital role in early diagnosis and subsequently, timely initiation of appropriate treatment. A multidisciplinary approach which includes pediatric dentist, orthodontist, oral surgeon and prosthodontist should be taken with an aim to provide the patient a better quality of life.

CONFLICT OF INTEREST & SOURCE OF FUNDING

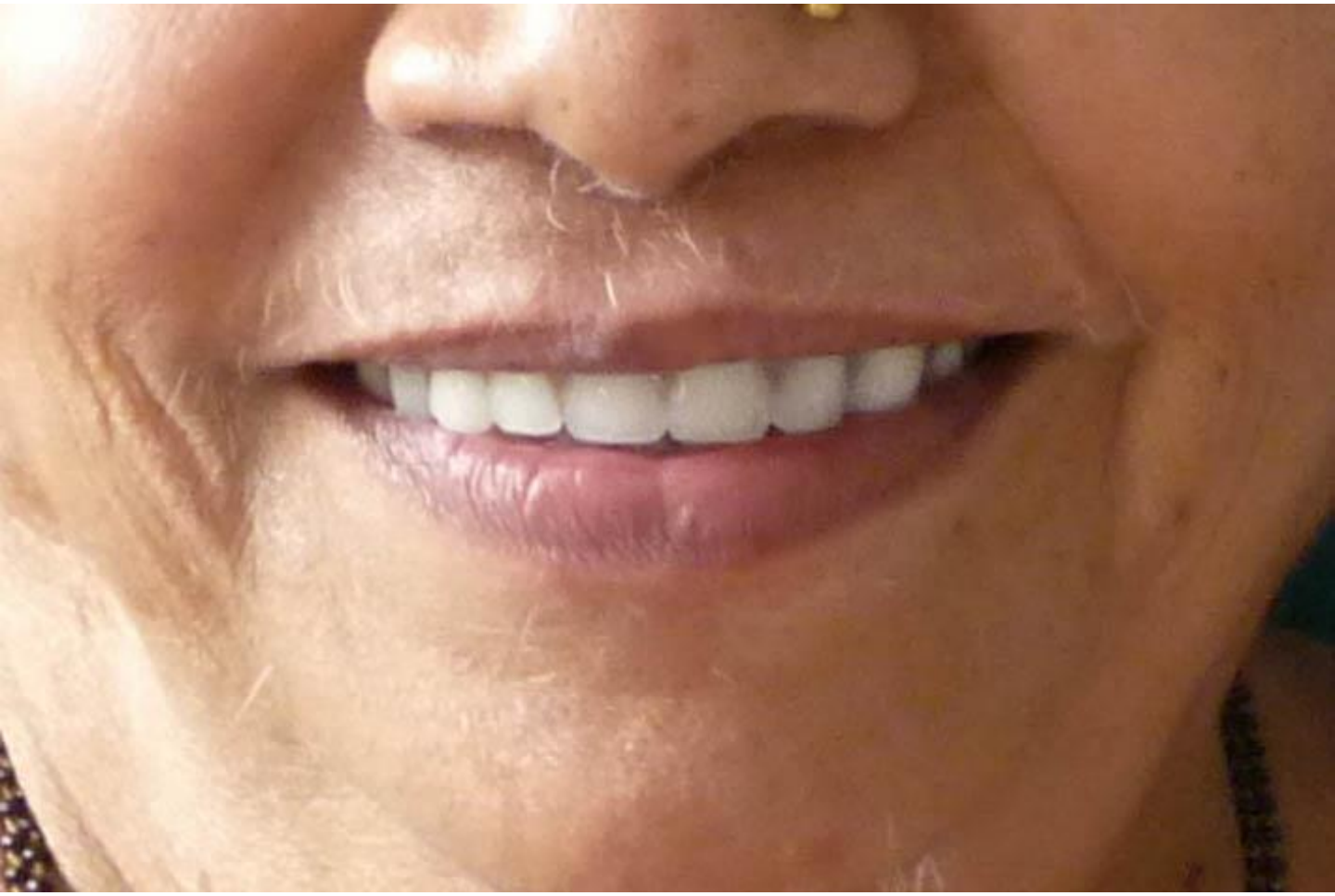
The author declares that there is no source of funding and there is no conflict of interest among all authors.

BIBLIOGRAPHY

1. 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):61-6.
2. Garg RK, Agrawal P. Clinical spectrum of cleidocranial dysplasia: a case report. Cases J 2008;1(1):377.
3. Feldman VB. Cleidocranial dysplasia-a case report. Journal of Can Chiropr Assoc 2002;46(3):185-9.
4. Bhat MA, Laway BA, Mantoo S, Choudry K, Kotwal S, Mir SA. Cleidocranial Dysplasia: a rare cause of disproportionate severe short stature. Journal of Medical Case Reports 2012;27(5):408-10.
5. Mundlos S. Cleidocranial dysplasia:clinical and molecular genetics. J Med Genet 1999;36(3):177-182.
6. Suresh SS. A family with cleidocranial dysplasia and crossed ectopic kidney in one child. Acta Orthop Belg 2009;75(4):521-7.
7. Karagüzel G, Aktürk FA, Okur E,

Gümele HR, Gedik Y, Okten A. Cleidocranial dysplasia - a case report. J Clin Res Pediatr Endocranol 2010;2(3):134-6.
8. Kolokitha OE, Ioannidou I. A 13 year old Caucasian boy with cleidocranial dysplasia: a case report. BMC Res Notes 2013;6:6.
9. White SC, Pharoh MJ. Oral Radiology-Principle and Interpretation. New Delhi: Elseiver; 2000.
10. Alves N, de Oliveira R. Cleidocranial dysplasia - A case report. Int J Morphol. 2008;26(4):1065-8.

Editorial Office
426/427, S. V. Road
B/30, 4th Floor
Roop Kamal Society
Kandivali (West)
Mumbai, Maharashtra
India (400067)



Volume 2 Issue 5 (Jul - Sep 2014) **ISSN 2347-6249**

International Journal of Oral Care & Research