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Dr. Radhika Sharma
Reader, Department of Oral Medicine and Radiology, GNDDC Sunam, Patiala, Punjab, India

Dr. Ashwarya Sharma
HOD and Professor, Department of Oral Medicine and Radiology, GNDDC Sunam, Patiala, Punjab, India

Dr. Zeeza Mehboob Qazi
MDS, Department of Oral Medicine and Radiology, GNDDC Sunam, Patiala, Punjab, India

Dr. Shruti Sharma
Professor, Department of Oral Medicine and Radiology, GNDDC Sunam, Patiala, Punjab, India

Dr. Puneet Goyal
Professor, Department of Paediatric and Preventive Dentistry, GNDDC Sunam, Patiala, Punjab, India

Dr. Sanna Choudhary
MDS, Department of Orthodontics and Dentofacial orthopaedic, GNDDC Sunam, Patiala, Punjab, India

Corresponding Author:
Dr. Radhika Sharma
Reader, Department of Oral Medicine and Radiology, GNDDC Sunam, Patiala, Punjab, India

Cleidocranial dysplasia: A rare case report

Dr. Radhika Sharma, Dr. Ashwarya Sharma, Dr. Zeeza Mehboob Qazi, Dr. Shruti Sharma, Dr. Puneet Goyal and Dr. Sanna Choudhary

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Abstract

Cleidocranial dysplasia (CD) is a rare autosomal dominant skeletal disorder, characterised by typical oral and skeletal disorders, the most striking feature of which is malformation or absence of clavicles, open anterior fontanelle, presence of wormian bones and multiple dental abnormalities such as delayed exfoliation / eruption or supernumerary teeth. Mostly such patients are first noticed by the dentist due to aesthetic reasons or delayed eruption of teeth. In this paper, a case of CD in a 16 year old female is reported and the literature is reviewed.

Keywords: Chromosome 6p21, clavicular defect, cleidocranial dysplasia, open anterior fontanelle, supernumerary teeth, runx2

Introduction

CCD is also known as Marie–Sainton disease, mutational dysostosis, and cleidocranial dysostosis. It is usually inherited in autosomal dominant manner and caused by mutation in human osteoblast – specific, runt-related transcription factor 2 (RUNX2) gene which is located on chromosome 6p21. Cleidocranial dysplasia (CCD) is a rare skeletal disorder which is characterized by delayed closure of anterior fontanelle, absent or hypoplastic clavicles and scoliosis of spine with short stature. Oral manifestations include delayed or failing eruption of permanent dentition with multiple supernumerary teeth, high arched or cleft palate, protruding mandible and mid face retrusion.

Case Report

A 16 year old female patient visited the department of Oral Medicine and Radiology with the chief complaint of unerupted permanent teeth in both upper and lower arch. There is history of discomfort while chewing food. Patient is born at term to nonconsanguineous couple, out of normal delivery. Patient's mother stated that patient was dull in grasping things and had learning disabilities. Her father had broad forehead with frontal bossing.

General physical examination revealed a moderately built and moderately nourished patient. On extraoral examination, the patient presented with broad forehead with frontal bossing, depressed nasal bridge, mid facial hypoplasia and flat face. (Figure 1,2) Wide open anterior fontanelle could be palpated in the anterior region of head. Shoulders appeared narrow and showed marked drooping. On palpation, clavicles were absent bilaterally and patient was able to approximate both the shoulders in the midline in the chest. (Figure 3) Intraoral examination revealed the presence of set of deciduous dentition with three permanent molars present being 51, 52, 53, 54, 16, 61, 62, 63, 64, 26, 72, 73, 75, 36, 82, 85 with carious w.r.t 16, 64, 26 and root stumps w.r.t 75 and a narrow high arched palate. (Figure 4, Figure 5).

Radiographic Examination

Orthopantomogram revealed multiple unerupted and supernumerary teeth. Unerupted teeth being 11, 12, 13, 14, 15, 17, 18, 21, 22, 23, 24, 25, 27, 28, 31, 32, 33, 34, 35, 36, 37, 38, 41, 42, 43, 44, 45, 46, 47, 48 with anomalous and distorted roots. Mandible demonstrated decreased trabeculation with reduced density in the mandible with flattened mandibular angle and pointed coronoid process. (Figure 6)

Paranasal sinus view revealed sinuses are poorly developed and open anterior frontanellae. (Figure 7)

Lateral cephalogram revealed a hypoplastic maxilla with multiple impacted teeth and incomplete development of mastoid air cells. (Figure 8)

Posteroanterior view of chest radiograph showed complete bilateral absence of clavicle. (Figure 9)



Fig 1: Frontal view with flat face, prominent forehead, sunken nasal bridge



Fig 2: Lateral profile view exhibiting midface deficit and concave profile



Fig 3: Physical stature with approximation of shoulder



Fig 4: Intraoperative view of mandible with multiple deciduous teeth and permanent tooth w.r.t 36



Fig 5: Intraoperative view of maxilla with multiple deciduous teeth and permanent teeth w.r.t 16, 26, with narrow and high arched palate



Fig 6: Panoramic view showing multiple unerupted and supernumerary teeth

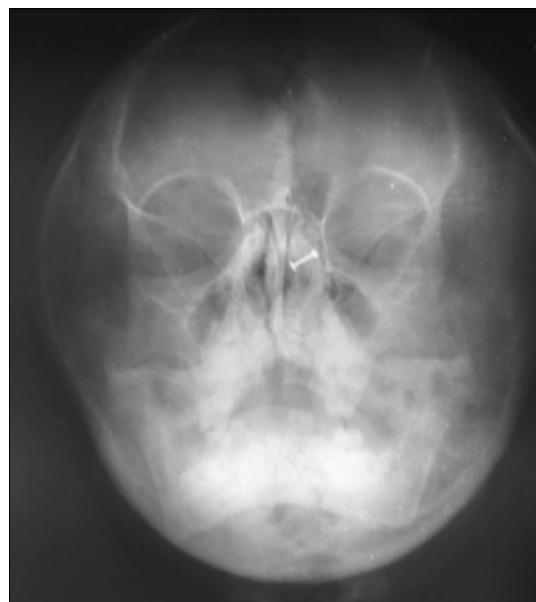


Fig 7: Paranasal sinus view exhibiting underdeveloped sinuses and open anterior fontanelle



Fig 8: Lateral cephalogram of skull showing hypoplastic maxilla and multiple unerupted teeth



Fig 9: Posteroanterior chest radiograph exhibiting clavicular hypoplasia

Based on the history taking and typical clinical examination a diagnosis of Cleidocranial dysplasia was made.

Discussion

Cleidocranial Dysplasia is a generalized skeletal dysplasia first described in 1765 by Martin and was termed as Cleidocranial dysostosis in 1897 by Marie and Sainton.

Synonyms

Marie Sainton disease, mutational dysostosis and cleidocranial dysostosis.

Etiology

This autosomal dominant disorder is caused by the mutation of the RUNX2 gene located on chromosome 6p21. The RUNX2 gene encodes various transcription factors essential for osteoblast differentiation. The mutated RUNX2 gene results in abnormal ossification of the affected endochondral and intramembranous bones. The disorder is characterized by several typical clinical and radiological findings. Although CCD is a congenital disorder, the spectrum of the clinical and radiological manifestations in the affected patients may vary considerably.

Clinical Features

- **Sex:** The disease affects men and women with equal frequency.
- **Sites:** It primarily affects skull, clavicle and dentition
- Shoulder meet in midline - A complete clavicle develops

from three different ossification centres. One or more of these ossification centers may be affected, resulting in clavicular hypoplasia/aplasia or discontinuous clavicles. All the three ossification centers are affected in only about 10% of cases and may result in complete clavicular agenesis. Partial or complete absence of the clavicle allows an exaggerated abnormal mobility at shoulders, enabling these individuals to approximate shoulders in the midline.

- **Head:** The head is brachycephalic (reduced anterior posterior dimension but increased skull width) or wide and short.
- **Skull:** In skull the fontanelles often remain open or at least exhibit delayed closing and for this reason, tend to be rather large. Open skull suture and multiple wormian bones are present and there is occasional stunting of long bone.
- **Lacrimal and zygomatic bone:** Lacrimal and zygomatic bone are also underdeveloped.
- **Sagittal suture:** Sagittal suture is characteristically sunken, giving skull flat appearance.
- **Nose:** Nasal Bridge is depressed with a broad base.

Oral Manifestations

- **Micrognathia:** maxilla and Paranasal sinus are underdeveloped, resulting in maxillary micrognathia.
- **Maxilla:** maxilla is underdeveloped and is smaller than normal, in relation to mandible.
- **Prognathism:** Maxilla is small and mandible is usually normal in size, which gives the appearance of prognathism.
- **Teeth:** Prolonged retention of primary dentition and delayed eruption of permanent dentition. Numerous unerupted teeth are found which are most prevalent in the mandibular, premolar and incisor area.
- **Palate:** High narrow arched palate and cleft palate may be common.
- **Hypoplasia of enamel:** The crown may be pitted as a result of enamel hypoplasia.

Radiographic Features

- **Skull finding:** skull film reveals open sutures, presence of wormian bones, widened cranium, delayed ossification of fontanelles, frontal and occipital bossing and basilar invagination.
- **Chest examination:** examination of chest reveals malformation and absence of clavicles.
- **Teeth:** jaw examination reveals prolonged retention of primary dentition. The unerupted teeth are frequently displaced to occupy an oblique or horizontal position in the jaw.
- **Supernumerary teeth:** presence of supernumerary teeth usually in anterior region. The supernumerary teeth in this area often show anomalous and distorted roots.
- **Jaws:** there is small underdeveloped jaw. There is also flattened mandibular angle and overgrowth of cranial base; anteroposteriorly and pointed coronoid process.
- **Roots:** roots of teeth are often somewhat short and thinner than the normal

Management

- Management of skull and clavicle anomalies—there is no treatment require for this anomalies. Patient can live its normal life.
- **Dental care:** as patient may be having supernumerary

teeth, they should be extracted. Prosthetic replacement of teeth should be carried out. Unerupted teeth should be extruded orthodontically. A multidisciplinary approach must be considered in the treatment of CD and a long time approach must be planned. The time of diagnosis of CD may also be suggested as a guide for the choice of the necessary treatment model.

Conclusion

Early detection is essential for prevention of morbidity and disability. Knowledge of the clinical characteristics, family history, and diagnostic tools for CD will enable the clinicians to achieve an early diagnosis and implement appropriate treatment to improve function and aesthetics. A holistic approach is required to take care of all the aspects, including the primary pathology and psychological aspects. Molecular genetics analysis can provide an absolute verification of the diagnosis of CD and allow for identification of the responsible gene.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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References

1. Chelvan HT, Malathi N, Kailasam V, Ponnudurai A. Cleidocranial dysplasia: A Family report. J Indian Soc Pedod Prev Dent. 2009;27:249-252. 10.4103/0970-4388.57661.
2. Golan I, Baumert U, Hrala BP, Müssig D. Early craniofacial signs of cleidocranial dysplasia. Int J Paediatr Dent. 2004;14:49-53.
3. Jensen BL, Kreiborg S. Craniofacial growth in cleidocranial dysplasia- a roentgenccephalometric study. J Craniofac Genet Dev Biol. 1995;15:35-43.
4. Jensen BL, Kreiborg S. Craniofacial abnormalities in 52 school-age and adult patients with cleidocranial dysplasia. J Craniofac Genet Dev Biol. 1993;13:98-108.
5. Kalliala E, Taskinen PJ. Cleidocranial dysostosis. Report of six typical cases and one atypical case. Oral Surg Oral Med Oral Pathol 1962;15:808-22.
6. Kolokitha OE, Papadopoulou AK. Cleidocranial dysplasia: Etiology, clinical characteristics, diagnostic information and treatment approach. Hell Orthod Rev. 2008;11:21-33.
7. Kreiborg S, Jensen BL, Larsen P, Schleidt DT, Darvann T. Anomalies of craniofacial skeleton and teeth in cleidocranial dysplasia. J Craniofac Genet Dev Biol. 1999;19:75-79.
8. McNamara CM, O'Riordan BC, Blake M, Sandy JR. Cleidocranial dysplasia: Radiological appearances on dental panoramic radiography. J Dento Maxillo Facial Radiol. 1999;28:87-89.
9. Mohan RP, Suma GN, Vashishth S, Goel S. Cleidocranial dysplasia: clinicoradiological illustration of a rare case. J Oral Sci. 2010;52:161-166.
10. Mundlos S, Otto F, Mundlos C, Mulliken JB, Aylsworth AS, Albright S, et al. Mutations involving the transcription factor CBFA1 cause cleidocranial dysplasia. Cell. 1997;89:773-9.
11. Roberts T, Stephen L, Beighton P. Cleidocranial dysplasia: A review of the dental, historical, and practical implications with an overview of the South African experience. Oral Surg Oral Med Oral Pathol Oral Radiol. 2013;115:46-55.
12. Suba Z, Balaton G, Gyulai-Gaál S, Balaton P, Barabás J, Tarján I. Cleidocranial dysplasia: Diagnostic criteria and combined treatment. J Craniofac Surg. 2005;16:1122-6.
13. Tanaka JLO, Ono E, Filho EM, Castilho JCM, Moraes LC, Moraes MEL. Cleidocranial dysplasia: importance of radiographic images in diagnosis of the condition. J Oral Sci. 2006;48:161-166. 10.2334/josnusd.48.161.
14. Xuan D, Li S, Zhang X, Lin L, Wang C, Zhang J. A novel RUNX2 mutation in cleidocranial dysplasia patients. Biochem Genet. 2008;46:702-707. 10.1007/s10528-008-9184-4.