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CLEIDOCRANIAL DYSPLASIA WITH *SPINA BIFIDA*: case report

Displasia cleido-craniana com espinha bífida: relato de caso

Mubeen Khan[a], Rai Puja[b]

[a] Professor and head of Department of Oral Medicine and Radiology Government Dental College and Research Institute, Bangalore - India.
[b] Postgraduate student, Department of Oral Medicine and Radiology, Government Dental College and Research Institute, Bangalore - India, e-mail: pujas.rai@gmail.com

Abstract

OBJECTIVE: To present and discuss a case of a rare disease in a 35 year old otherwise healthy male Indian in origin reported to the Department of Oral Medicine and Radiology of the Dental College and Research Institute, Bangalore, India. **DISCUSSION**: The cleidocranial dysplasia is a rare disease which can occur either spontaneously (40%) or by an autosomal dominant inheritance. The dentists are, most of the times, the first professionals who patients look for to solve their problem, since there is a delay in the eruption and /or absence of permanent teeth. In the present case multiple missing teeth was the reason for patient's visit to odontologist. **CONCLUSION**: An early diagnosis allows proper orientation for the treatment, offering a better life quality for the patient.

Keywords: Cleidocranial dysplasia. Aplastic clavicles. Delayed eruption. Supernumerary teeth. *Spina bifida*.

Resumo

***OBJETIVO**: Apresentar e discutir um caso de doença rara em paciente masculino, de 35 anos de idade, sadio, de modo geral, de origem indiana, que foi encaminhado ao Departamento de Medicina Bucal e Radiologia da Escola de Odontologia e Instituto de Pesquisa, Bangalore, Índia. **DISCUSSÃO**: A displasia cleidocraniana é uma doença rara que pode ocorrer espontaneamente (40%) ou por herança autossômica dominante. O cirurgião-dentista é, na maioria das vezes, o primeiro profissional que o paciente procura para solução de seu problema, uma vez que há demora na*

erupção e/ou ausências de dentes permanentes. No presente caso, múltiplos dentes ausentes foram a razão para o paciente visitar o dentista. ***CONCLUSÃO****: O diagnóstico precoce permite a orientação adequada para o tratamento, visando melhor qualidade de vida ao paciente.*

Palavras-chave*: Displasia cleido-craniana. Clavículas aplásticas. Erupção retardada. Dentes supranumerários.* Spina bifida.

INTRODUCTION

The cleidocranial dysplasia (CCD), also known as Marie and Sainton disease, Scheuthauer Marie-Sainton syndrome and mutational dysostosis, is a rare inherited skeletal dysplasia with incidence of 1:100,0000 (1). Cleidocranial dysplasia is a dominant, inherited autosomal bone disorder with a wide range of expressivities, primarily affecting bones undergoing intramembranous ossification (2). The CCD gene has been mapped to chromosome *6 p21* within a region containing the *CBFA1* gene, a member of the Runt family of transcription factors (3), which controls differentiation of precursor cells into osteoblasts. It is thus essential for membranous as well as endochondral bone formation, which may be related to delayed ossification of the skull, teeth, pelvis and extremities in CCD (4, 5). The genotype–phenotype correlations in mutational studies of the *RUNX2* domain show a variable clinical spectrum, suggesting that skeletal growth and dental development could be related to the type of changes in the *RUNX2* activity (6).

Cleidocranial dysplasia is characterised by disturbances in the growth of bones involving cranial vault, clavicles, maxilla, nasal bones, lacrimal bone, zygomatic bone, vertebrae and the pelvis (7). Spina bifida, however, remains as rare finding in this syndrome with very few reports in the literature. Patients with CCD usually present with short stature, frontal, parietal and occipital bossing with underdeveloped paranasal *sinus*. Hypertelorism with the bridge of the nose appearing wide and flat, underdeveloped maxilla with relative mandibular prognathism are common. The ability to approximate the shoulders anteriorly is related to clavicular hypoplasia/ absence and is the classic diagnostic sign of the disorder (7). Dental problems present the most significant manifestation of CCD; they usually include retention of multiple deciduous teeth, impaction or delay in eruption of permanent teeth, presence of a varying number of supernumerary teeth, an increase in the osseous density of some regions as a result of an abnormal remodelling of the bone and absence or less quantity of the cellular cementum in the roots of the permanent/ deciduous teeth (8).

Dental abnormalities are sometimes the sole clinical signs of the syndrome. Due to the wide variability and subclinical presentation in many patients, diagnosis may be difficult. Since early diagnosis of CCD is essential for initiating the appropriate treatment approach, physicians should be aware of the characteristic features of this syndrome. A detailed review regarding the entire clinical and radiographic picture does not exist. This paper aims at discussing clinical and radiographic features of cleidocranial dysplasia along with a rare finding of *spina bifida*.

Case report

Thirty five year old otherwise healthy male Indian in origin reported to the Department of Oral Medicine and Radiology, with a chief complaint of congenitally missing teeth and desired there replacement. This was the patient's first visit to health care professionals. Upon general examination, he was well oriented, of average height and normal gait. The skull was brachycephalic with frontal, parietal and occipital bossing. Patient had hypertelorism, with the bridge of the nose appearing wide and flat.The midface was depressed because of underdeveloped maxilla, leading to mandibular pseudoprognathism giving patient a concave facial profile. Bilateral zygomatic bones were hypoplastic leading to flattening of checks. Patient also had broad neck, sloping and hyper mobility of shoulders with tendency to approximate them anteriorly close to mid line (Figure 1).

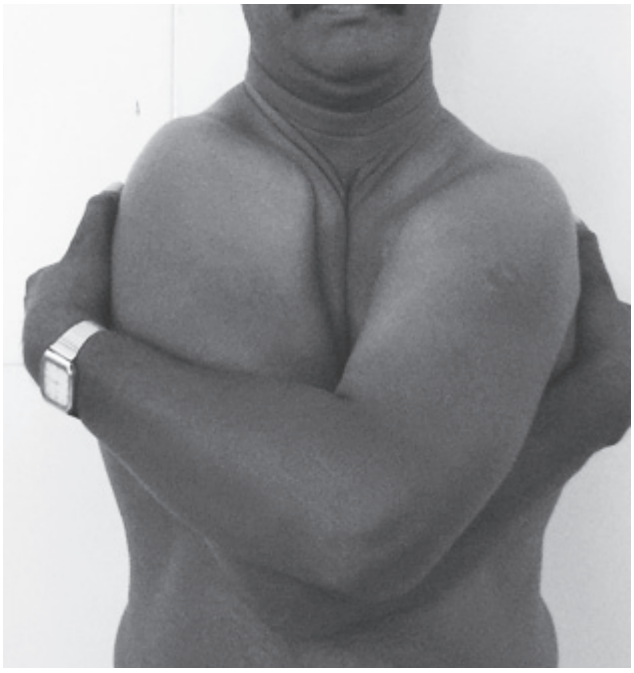

Figure 1 - Patient's frontal view

Intra oral examination revealed presence of the following teeth:

7 6 5 4 2 1	1 2 D E 6 7
7 E D C	E 7

with clinically missing

3	3 4 5
6 5 4 3 2 1	1 2 3 4 5 6

which were impacted. Two supernumerary teeth were present in maxilla and mandible respectively. A deep high arched narrow palate was present (Figure 2).

The oral hygiene was generally poor. The oral mucosa and tongue were otherwise normal. Family history revealed similar signs in the mother; his spouse and offspring were normal. Based on familiar history and clinical features, a provisional diagnosis of CCD was given. A differential diagnosis of Noonan syndrome, Turner's syndrome, rickets, *osteogenesis imperfecta*, and Gardner´s syndrome was given. The patient was subjected to radiographic examination. Orthopantomogram revealed multiple unerupted permanent teeth and impacted supernumerary teeth in both the upper and lower jaws. The mandibular notch showed excessive deepening causing elongated appearance of coronoid process. The gonial angle appeared rounded with lack of definition of the neck of condyle (Figure 3).

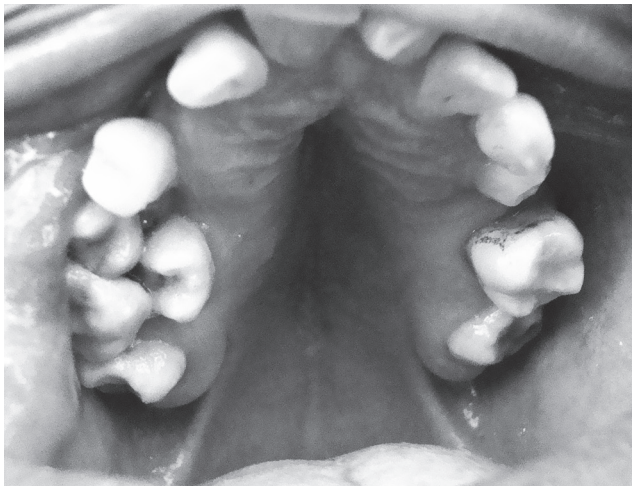

Figure 2 - Intra oral view

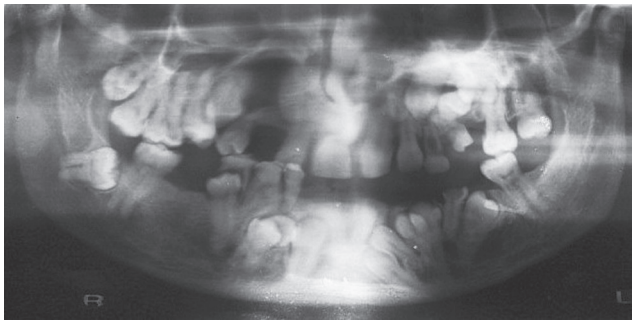

Figure 3 - Orthopantomogram

The postero-anterior view and laleral skull view showed a brachycephalic skull, noticeable frontal, parietal and occipital eminences. The anterior and posterior *fontanelles* were widely opened with the suture shadows excessively prominent. The maxilla and zygomatic bones were relatively small with underdeveloped frontal and maxillary air sinus (Figures 4, 5).

X-ray of the chest showed absence of both clavicles and narrowing of the thoracic cage. Vertical radiolucencies noted in the spinous process of C_7 to T_3 vertebrae suggestive of *spina bifida* (Figure 6).

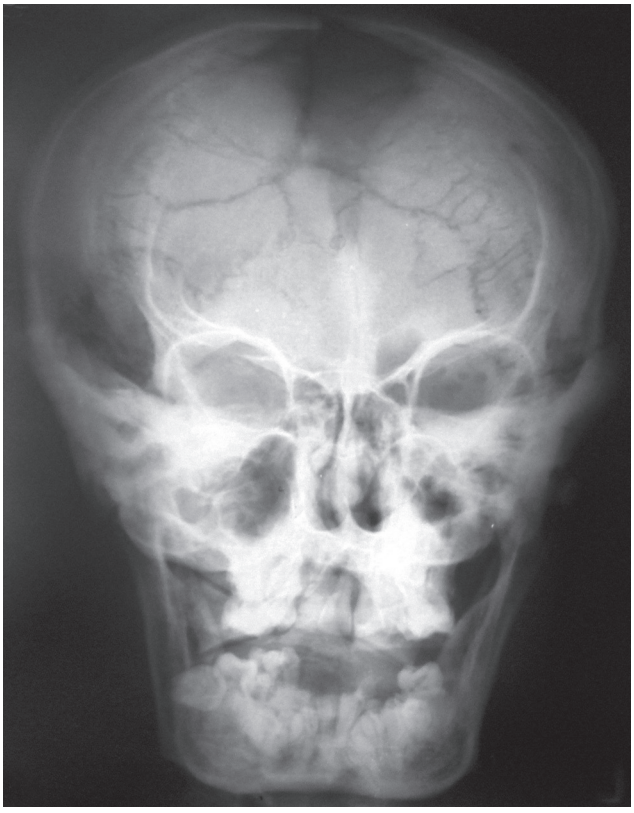

Figure 4 - Frontal skull view

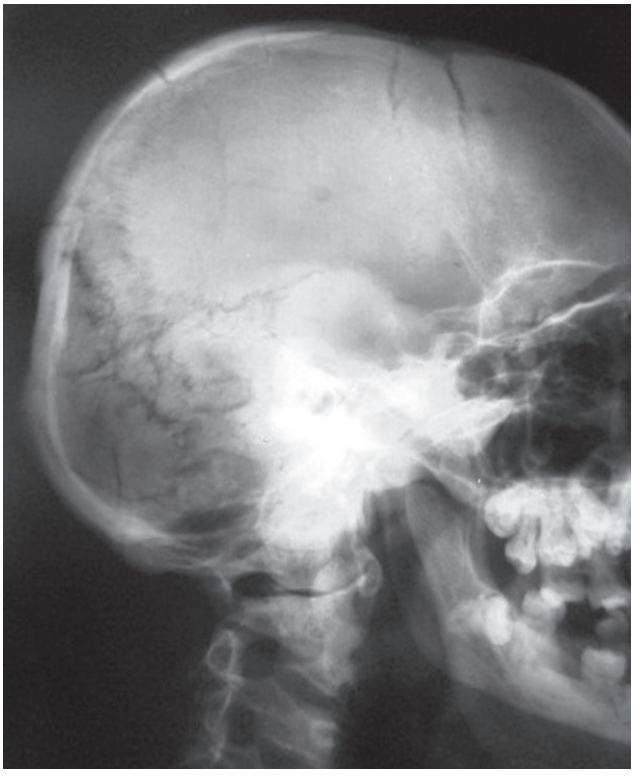

Figure 5 - Lateral skull view

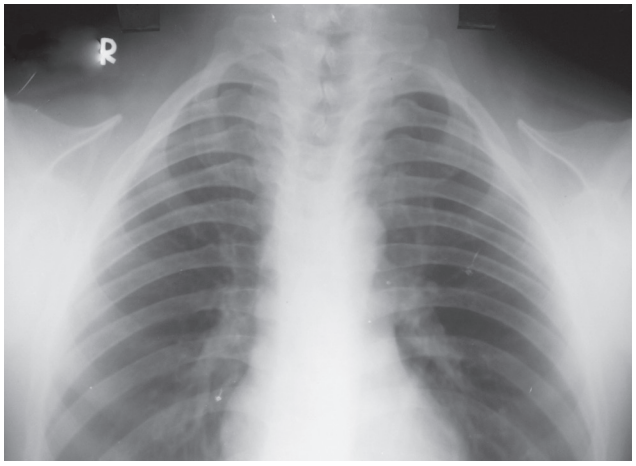

Figure 6 - Radiolucencies in the spinous process of C_7 to T_3 (*spina bifida*)

On the basis of the familiar history, clinical features and radiographic findings, a final diagnosis of cleidocranial dysplasia with *spina bifida* at the level of C_7 to T_3 vertebrae was given.

DISCUSSION

In 1760, Meckel recorded an infant born without clavicles (9). Marie and Sainton described four cases in detail and gave the condition its name of "hereditary cleidocranial dysostosis" (10). The terminology has changed from dysostosis – meaning abnormal bone formation – to dysplasia – meaning abnormal tissue development, thereby reflecting the more generalized nature of the disease process (11). There is a generalized failure of midline ossification resulting in patent *fontanelles*, metopic suture, wormian bones, nasal deformity, non-union of mandibular symphysis, high arched palate, cleft palate, hypoplasia or absence of clavicles, *spina bifida* and delayed closure of pubic symphysis (12). Depression of the sagittal suture has been described as the "hot cross bun effect". Calvarial thickening may be present affecting the squamous portion of the temporal bone or the occipital bone. The most characteristic and pathognomic skeletal feature is that one or both clavicles are frequently partially or in 10% cases completely absent. Clavicle being first bone to be ossified in the 6th week of fetal life is most often affected (13). Clavicular deformity along with the dysplastic muscle attachments give rise to elongated

neck; narrowness, drooping and hypermobility of shoulders with tendency to approximate shoulders anteriorly. All these features were observed in the present case. Reported vertebral abnormalities are scoliosis, kyphosis, lordosis and vertebral synostosis. Hearing loss and complications during child birth and dislocation of joints can be present. Although psychosocial disorders associated with abnormal facial and body features may occur, patients have normal intelligence, with an overall good prognosis and normal life expectancy (13).

Regarding the manifestations of dentistryinterest it is important to mention that these patients show an arcate, narrow and deep palate. The maxilla can be underdeveloped and shorter than normal in relation to the mandible, resulting in a pseudo mandibular prognathism. Underdeveloped zygomatic and lacrimal bones can also be present. Hypoplastic paranasal sinuses have been reported. Another important finding is the presence of supernumerary teeth that forms as a result of activation of remnants of the dental lamina left unresorbed during odontogenesis. Crowding of the dental arches caused by these supernumerary teeth may play a role in arresting the eruption of permanent teeth or forcing them into ectopic locations. The dental eruption is retarded and an absence of root resorption in the deciduous teeth, hypodontia and dentigerous cysts can be observed. It has been reported that masseter muscle size, bite-force magnitude, and craniofacial bone shape and structure are mutually related hence a low bite force & less thick masseter muscle has been reported in CCD patients (14).

Treatment of the dental problems associated with cleidocranial dysplasia may be difficult and requires an interdisciplinary approach. Therapeutic options include extraction of all teeth followed by the fabrication of dentures or a crown sleeve coping overdenture, autotransplantation (15) of selected impacted teeth followed by prosthetic restoration or removal of primary and supernumerary teeth followed by exposure of permanent teeth that are subsequently extruded orthodontically. The use of implants in a patient with cleidocranial dysplasia to support a removable overdenture has been documented (16).

Early diagnosis of CCD is essential for appropriate treatment approach based on interdisciplinary cooperation between the dentist at the right moment, offering a better quality of life for the patient.

CONSENT FORM

The authors declared that the patient signed a consent form for publication of this case and that the publication was approved by the Ethical Comittee of the Institution.

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