

Cleidocranial Dysostosis: Report of Case from Saudi Population with New Radiographic Finding in Infraorbital Rim

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Abstract Cleidocranial dysplasia (CCD) is a rare autosomal dominant genetic disorder which is characterized by its unique intraoral and extra features. Sometimes the disease occurs spontaneously. Here we describe a case of spontaneous occurrence from Saudi Arabia due to its extreme rarity in this continent.

Keywords: *Cleidocranial dysplasia, frontal bossing, clavicle, brachycephaly, Runx2 Gene*

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1. Introduction

Cleidocranial dysplasia is a rare genetic disorder which manifests with wide variability. It is characterized by hypoplastic or aplastic clavicles, short stature, by persistently open or delayed closure of the fontanel, and dental anomalies. [1,2,3,4] This may be associated with skeletal deformities. [5] Usually the dentists are the first to encounter these cases during the routine oral examination. It is important for dentists to fully understand the clinical manifestations and its variations. This case report is important that only one case from Saudi population has been reported. This may be due to genetic features among the Saudi population. Thus we are reporting this case due to its rarity in the Saudi population and add one more case for demographic purposes.

2. Case Report

A 36 year-old male attended to our hospital with the chief complaint of inability to chew food due to bad teeth. The history revealed that the whatever teeth were present, his many teeth were extracted due to different reasons and most of them were milk teeth and the last tooth was extracted last year only. His main problem was difficulty in chewing due to absence of teeth. Intraoral examination revealed poor oral hygiene, multiple carious and hypoplastic teeth, retained primary teeth, and absence of permanent teeth. The extra-oral examination showed hypertelorism, cranial bossing and he could not bring his shoulders closer together (Figure 1). On the basis of clinical findings the case was provisionally diagnosed as a case of

Cleidocranial Dysplasia (CCD) which was later confirmed by radiographic examination. PA view of chest revealed partial absence of clavicle. (Figure 2). In the panoramic radiograph there were many impacted teeth but were palpable, many root stumps (Figure 3).

A rare finding in this case we deciphered in CT image (Figure 4c) as bilateral radiolucent intrabony lesion on infraorbital floor, without any correlation with pathological lesion.

Further, ancestral analysis was impossible as the grandparents had passed away. However among brothers, sisters, uncles of patient no history of any genetic disorders could be traced. His children were also disease free from any genetic disease.



Figure 1. Profile photograph with shoulders brought closer



Figure 2. PA View chest. Bilaterally partially developed clavicles



Figure 3 a. Orthopantomogram showing impacted and supernumerary teeth

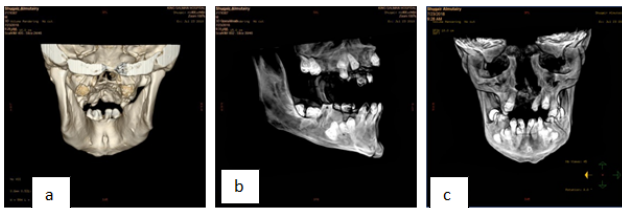


Figure 4. A, b CT images showing dental status and c showing poorly developed infraorbital floor with bilateral radiolucent intrabony lesion on infraorbital region and supernumerary teeth

3. Discussion

Cleidocranial dysplasia has been considered as a genetic disorder and in most of the cases a familial incidence has been reported. It is seen approximately 33% of the reported cases and its occurrence has been reported in as many as five successive generations. The disease appears to be inherited as autosomal dominant disease. [6] However, chances of sporadic occurrence can't be ignored and our case may be considered as an example of that. The family history in this case did not reveal any affliction in direct ancestors or descendants. The family history could be traced up to two generations, it did not reveal any presence of such disease. In such cases the disease is possibly inherited as recessive character or new dominant mutation or as an incomplete change in a genetic trait with different gene expression. [7]

This case presents many extra oral and intra oral characteristic anomalies which are characteristic of CCD. Individuals with this disorder bear characteristic features like reduced height, large brachycephalic head, small and angular face, prominent frontal and parietal bones and drooping shoulder with excessive mobility. Clavicular involvement may vary and depending on the amount of its involvement. [8] Intraoral findings are high arched palate, retention of deciduous teeth, delayed eruption of

permanent teeth and the presence of large number of impacted supernumerary teeth. [9] Mid line deformities are very common due to generalized failure of midline ossification. Thus this leads to patent fontanella, metopic suture and wormian bones, nasal deformity, hypoplasia or absence of clavicles, spina bifida and delayed closure of pubic symphysis. Oral deformities may be manifested as non-union of mandibular symphysis, high palatal vault, cleft palate. [10] These all the features were present in our case. In this case we observed a so not reported finding i.e. bilateral intrabony cystic appearance in infra orbital rim, without any clinical or pathological association.

Previous studies have indicated in 2% cases, the height of the individual remains normal up to age of 4 to 8 years. [11] In addition there is some disproportionate short stature with short limbs compared to trunk, more apparent in upper limbs than in lower limbs. Although literature reveals more affliction in females but previous case report from Saudi Arabia is from patient and this is second case. Due to rarity of this disease in Saudi Arabia its predilection in Females could not be confirmed. [5] Other peripheral abnormalities include delayed mineralization of pubic bone, broad pubic symphysis, and narrow pelvis. Occasionally spina bifida may be also present and thoracic cage may be small and bell shaped with short ribs. [12,13]

4. Conclusion

In individuals suffering from CCD, certain complications are common which include dental caries, osteomyelitis of the mandible or maxilla, respiratory distress in early infancy, in late stages shoulder and hip dislocation, upper airway complications, recurrent ear infection, hearing loss. [14,15]

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