Cleidocranial Dysplasia in Son and Father: Report of Two Rare Cases with Review of Literature

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ABSTRACT

Cleidocranial dysplasia (CCD) is a rare autosomal dominant skeletal disease, which is caused by mutation in the gene on 6p21 encoding transcription factor CBFA1, i.e. runt-related transcription factor 2 (RUNX2). The disease is characterized by a persistently open anterior fontanelle and skull sutures, hypoplastic or aplastic clavicles, dental abnormalities, short stature, a wide pubic symphysis and a variety of other skeletal changes. A major finding of CCD is hypoplasia or aplasia of clavicular bones resulting in the ability of the patient to approximate the shoulders. Delayed closure of the anterior fontanelle and of metopic sutures causes frontal bossing. We report a rare case of CCD in a 15-year-old boy and in his father and emphasize the importance of clinical and radiographic findings in CCD.

Keywords: Cleidocranial dysplasia, Unclosed fontanelle, Frontal bossing, Aplasia of clavicle.

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INTRODUCTION

Cleidocranial dysplasia (CCD) is a rare dominantly inherited autosomal bone disease that 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. Delayed closure of the anterior fontanelle and metopic sutures results in frontal bossing. The phenotypic spectrum ranges from mildly affected individuals with dental anomalies only to severely affected patients with syringomyelia.\textsuperscript{1,2} CCD is also known as Marie-Sainton disease, mutational dysostosis and cleidocranial dysostosis.\textsuperscript{3} Human osteoblast-specific, runt-related transcription factor 2 (RUNX2) gene located on chromosome 6p21 is identified as the gene responsible for CCD.\textsuperscript{4}

Here, we report a rare case of CCD in a 15-year-old boy and in his father and emphasize the importance of clinical examination and radiographic findings.

CASE REPORT

Patient named Mahendran aged 15 years came to Ragas Dental College and Hospital OP with a chief complaint of failure of eruption of teeth in upper and lower front tooth region for the past 5 years. The patient had normal exfoliation of teeth by 6 to 10 years of age, after which the permanent teeth had not erupted till date (Fig. 1).

The patient had consulted various dental practitioners for the same complaint.

Family history revealed that only his father had a similar complaint of retained deciduous teeth and some unerupted permanent teeth.

Height was 145 cm and weight was 35 Kg. Forehead revealed brachycephaly, biparietal and frontal bossing was present. A groove is present in the forehead which extends up to the metopic suture.

Fig. 1: Intraoral view showing numerous overretained deciduous and some unerupted permanent teeth in son and father.
Facial appearance revealed hypertelorism, depressed nasal bridge with a broad nasal base. Ears are smaller in size in comparison to normal individuals.

Hyperextensibility was present in the shoulders and patient was able to approximate the shoulders in front of his chest. The above features were present in patient’s father also (Fig. 2).

RADIOGRAPHIC FINDINGS

OPG showed the presence of multiple, unerupted crowns portion of tooth below the roots of primary teeth in relation to 51 to 55 in son and some unerupted teeth in father (Fig. 3).

PA skull revealed the presence of delayed closure of frontal and sagittal sutures in both father and son (Fig. 4).

Lateral skull showed numerous wormian bones present along the lateral part of frontal and sagittal suture lines. It also revealed a hypoplastic maxilla with a deficient zygomatic bone which gives the appearance of prognathic mandible in both father and son (Fig. 5).

PA chest showed partial absence of clavicles on the right and left sides in both father and son (Fig. 6).

Right and left hand wrist radiographs showed long second metacarpals and cone-shaped epiphyses on the distal phalanges on the right and left hand in both father and son (Fig. 7).

Pelvis AP showed delayed closure of pubic symphysis in both father and son (Fig. 8).

Considering the above clinical and radiographic features, a final diagnosis of CCD was given.

DISCUSSION

The major features of CCD are aplastic or hypoplastic clavicles, dental abnormalities (multiple supernumerary teeth, multiple impacted permanent teeth, retention of the deciduous teeth) and delayed closure of the sagittal fontanelles. Typically, our patient and his father had all of these findings that are pathognomonic for a diagnosis of CCD. Other findings of CCD are short stature, a bell-shaped thorax, hypoplasia of the pelvis, enlargement of the frontal and occipital bones and phalangeal abnormalities. Shortened or absent nasal bones, paranasal sinus abnormalities, thickening of some segments of the calvaria, small maxillae, and delayed union of the mandibular symphysis are less common findings of CCD. There is a notably phenotypic variation of CCD even within one and the same family. In approximately 40% of CCD patients, a genetic transition cannot be identified and the condition develops spontaneously.1,5-7

Fig. 2: Facial profile view of the patient and father demonstrating hypermobility of the shoulder girdles and frontal bosselation

Fig. 3: Panoramic view of the jaws showing multiple unerupted supernumerary teeth mimicking premolar, missing gonial angles and underdeveloped maxillary sinuses in son and father

Fig. 4: PA skull revealed the presence of delayed closure of frontal and sagittal sutures, multiple wormian bones in son and father
Clavicles are underdeveloped to varying degrees in these patients and are completely absent in approximately 10%. This allows excessive mobility of the shoulder girdle, as was also observed in our patient and in his father.

Dental abnormality is one of the main features of CCD. Our patient had retained deciduous teeth and multiple supernumerary teeth, which can impede the normal eruption of permanent teeth. It has been suggested that supernumerary teeth in such cases should be removed as soon as possible.6,8

The main finding in our patient and in his father was an open anterior fontanelle. Delayed closure of fontanelles could be a feature of hypothyroidism, rickets, hypophosphatasia, osteogenesis imperfecta, pycnodysostosis and other syndromes, such as Apert syndrome, Dubowitz syndrome, Russell-Silver syndrome, Down’s syndrome and Crouzon syndrome.9 When other characteristic features are taken into account, CCD can be differentiated easily from the other skeletal disorders and syndromes.

It is known that CCD is caused by heterozygous mutations in RUNX2 gene, which encodes a transcription factor required for osteoblast differentiation and is located on chromosome 6p21.1,4 Many mutations in the RUNX2 gene have been identified in patients with CCD.

The suggested treatment for dental complications of cleidocranial dysplasia is:
1. Fabrication of dentures over the unerupted teeth, and
2. Removal of teeth as they erupt, for very little bone structure would be left if supernumerary, impacted and unerupted teeth were all extracted at once.7,10
CONCLUSION
The clinical findings of cleidocranial dysplasia, although present at birth, are often either missed or diagnosed at a much later time. Some cases are diagnosed through incidental findings by physicians, treating patients for unrelated conditions. CCD may be identified by family history, excessive mobility of shoulders and radiographic pathognomonic findings of the chest, skull and jaws.

REFERENCES

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