CASE REPORT

CLEIDOCRANIAL DYSPLASIA : A REPORT

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ABSTRACT

Cleidocranial dysplasia is a rare disease also known as Marie and Sainton disease, and mutational dysostosis. The disease is transmitted by an autosomal dominant mode of inheritance with high penetrance and variable expressivity. A spontaneous mutation occurs in nearly one third of cases and mutation in the core binding factor α 1 (CDFA-1) gene, located on chromosome 6p21 has been reported. The condition is characterized by open sutures of skull bones, hypoplastic clavicle, multiple supernumerary teeth and unerupted permanent teeth of normal series. Here we have described a 14 year old boy with cleidocranial dysplasia. The patient had classical features of this disease.

KEY WORDS

Cleidocranial dysplasia, multiple supernumerary teeth, unerupted permanent teeth, retained primary teeth

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INTRODUCTION

Cleidocranial dysplasia (CCD, OMIM 119600) is a rare autosomal dominant disorder occurring one in million live birth, equally affecting both sexes. The disease is also known as Marie Sainton's disease. Initially the term 'cleidocranial dysostosis' was used because; it was believed that only bones of intra membranous origin were involved. However, it is now established that bones of endochondral ossification are also affected, thus a more generalized term 'cleidocranial dysplasia' is used.[1] The bony disorder is caused by mutation in the core binding factor Alpha-1 gene (CBFA1) of chromosome 6p21. The CBFA1 gene controls osteoblastic differentiation, thus playing important role in membraneous, as well as endrochondral bone formation.[2-4] Recent studies have indicated that CBFA 1 gene also plays an important role in tooth development through participation in odontoblast differentiation.[2] Although the disease shows an autosomal dominant pattern of inheritance, in 40% of cases spontaneous mutations occur.[1-5] The phenotypic expression of CCD shows wide variations, even among family members. The purpose of this article is to describe a case of CCD that was not diagnosed before.

CASE REPORT

A 14 year old boy reported with tooth ache. The boy had a normal intelligence and short stature for his age. [Figure 1] Physical examination revealed, frontal bossing, prominent forehead, depressed nasal bridge, hypertelorism, maxillary hypoplasia, and prognathic mandible. His shoulders were hypermobile. His family history and past medical history were noncontributory. Parents reported that his primary teeth had erupted in time. The intra oral examination showed a high arched palate with a mixed dentition stage. [Figure 2] The patient had primary maxillary incisors, all primary canines, permanent mandibular incisors, all premolars, permanent first molars and second molars. [Figure 3] Attrition of maxillary primary incisors was observed.

The antero-posterior and lateral skull

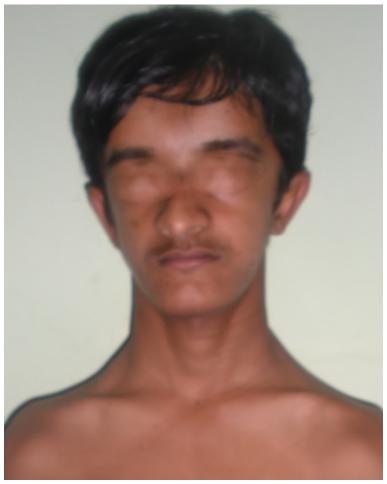

Figure 1. Photograph showing appearance of the patient.

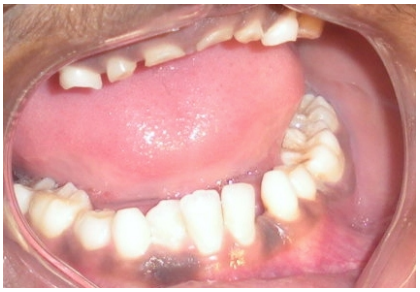

Figure2. Patient with high arched palate.

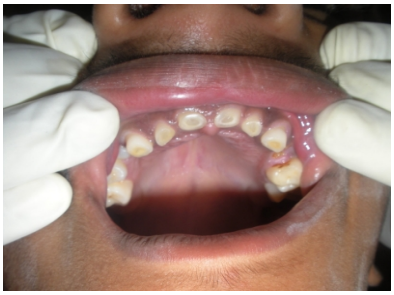

Figure3.Intra oral photograph showing a mixed dentition stage with severe attrition of maxillary primary anterior teeth.

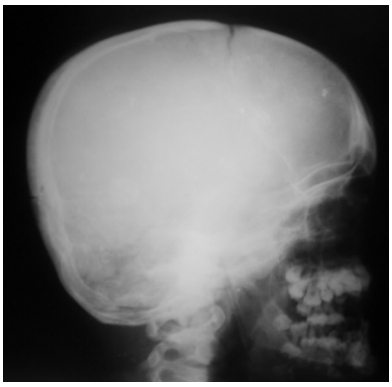

Figure 4. Xray lateral skull showing open anterior fontalelli.

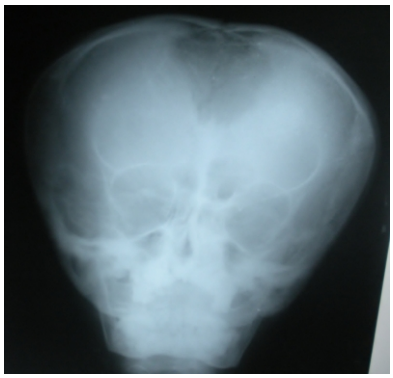

Figure 5. Antero posterior view of skull showing open fontanelli.

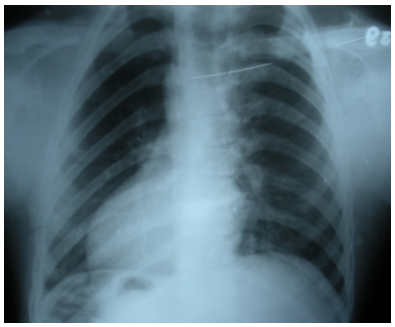

Figure 6. Bell shaped chest

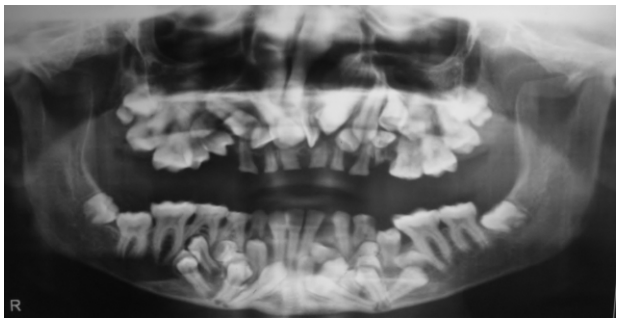

Figure 7. OPG showing multiple supernumeraries and unerupted teeth.

radiograph exhibited open anterior fontanels [Figure 4 & 5] Radiographic examination also showed a bell shaped chest with clavicular hypoplasia. [Figure 6] Panoramic examination displayed over retention of primary teeth, unerupted permanent teeth of normal series, and multiple supernumerary teeth. [Figure 7] Nearly parallel anterior and posterior borders of ascending ramus and distinctive curve of sigmoid notch were also observed. Based on clinical and radiological features, a diagnosis of CCD was made.

All retained primary teeth were extracted in multiple appointments and the patient was kept under observation.

DISCUSSION

The characteristic features of CCD are hypoplasia or aplasia of clavicle, delayed closure of cranial sutures, and dental abnormalities including prolonged retention of primary teeth, failure of eruption of permanent teeth, and presence of supernumerary teeth. Our patient had all these features.

One of the important features of this bony disorder is clavicular hypoplasia. Although clavicles may be absent in 10% of cases, but more commonly, acromial end is deficient.[4,5] Clavicular hypoplasia

leads to unusual shoulder mobility, and in some cases, both shoulders can be anteriorly approximated. Our patient displayed hyper mobility of shoulders. Abnormalities of skull include, open fontanels, presence of woormian bones in cranial sutures, and bossing of frontal, parietal, and occipital bones, small or absent nasal bones. Additionally, maxillary hypoplasia, high arched palate, and mandibular prognathism are also observed in CCD patients. [1,3,5]

Skeletal abnormalities of other bones include scoliosis, and kyphosis of spines, delayed ossification of pubic bones, bell shaped thorax, short and oblique ribs, short middle phalanges, hypoplastic distal phalanges of hands. [1]

Primary teeth in CCD patients tend to get over retained. Impaction of permanent teeth and multiple supernumerary teeth are also common dental features of CCD. Multiple supernumerary teeth may lead to dental crowding and failure of eruption of permanent teeth. Failure of eruption may also be due to other factors like, lack of cellular cementum, abnormal resorption pattern of alveolar bone, and increased bone density.[5] Despite a variety of skeletal problems, systemic effect of CCD is often mild and requires no treatment. However, dental problems including esthetics and functional problems like malocclusion may need professional attention. And many a times a patient reports to a dentist to correct his or her dental problems. The clinical and radiographic examinations of oral cavity constitute an important part diagnosis of this rare disorder.

Regarding the dental management of CCD, earlier the treatment is initiated, better is the prognosis.[4] In our patient, painful primary maxillary incisors were extracted. Management option for retained primary teeth, and supernumerary teeth is also surgical removal to facilitate eruption of impacted permanent teeth. However, many cases require surgical exposure, and orthodontic traction to bring impacted permanent teeth in to their esthetically and functionally acceptable position.[5,8] In our case, all primary teeth were extracted, and the patient was kept under observation.

One of the striking features of CCD is clavicular hypoplasia. But this abnormality is also seen in pyknodysostosis, mandibuloacral dysplasia, and Yunis Varon syndrome.[1,4,5] Pyknodysostosis has many features common with CCD. However, absence of supernumerary teeth, and increased bone density make differentiation easier from CCD. Mandibuloacral dysplasia is characterized by acroosteolysis and progressive stiffening of bones. Intellectual dysfunction is a feature of Yunis Varon syndrome.

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