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Case Report

A Case of Cleidocranial Dysplasia with Historical and Current Review

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Abstract: ***Introduction:*** This article discusses about our case of cleidocranial dysplasia and a review for dental manifestation and management of these cases. This disorder has a genetic background and general manifestations of CCD are outlined and an overview is presented. ***History:*** CCD is one of the best studies and documented disease whose history dates back to the prehistoric times with first case documented by Greig in 1933 in the museum of royal college of surgeons in Edinburgh. In the year of 1871, Scheuthauer published cranial and non – cranial findings of CCD. ***Case Report:*** We present a case of a 17 year old male patient who reported to our hospital with a complain of smaller and yellowish discolored teeth with spacing in the arch causing difficulty in chewing food. We have documented this case to the best of our understanding and with pictures depicting the patient and his clinical and radiographic findings. An interesting overview of the patient has been described. ***Discussion:*** We have researched different articles related to our patient and have documented the prominent features seen in these patients and correlated with the findings in our patient. There are reports of multiple patients who were diagnosed at a much earlier stage than our patient. Most of the clinical features described in the previous literature coincide with other findings, however additional clinical findings have been mentioned irrespective of their presence in our patient or not. ***Conclusion:*** We conclude that a proper clinical and radiographic evaluation can lead to early diagnosis of these patients and hence can aid in their management and education of the patient and his care takers. There are no reported cases of deficient IQ or abnormal life experienced by these patients except for that related to short stature.

Keywords: Cleido cranial dysplasia, Cleidocranial dysostosis, CCD, Autosomal recessive CCD.

INTRODUCTION

CCD is a genetic skeletal dysplasia in which hypoplasia of clavicles and deficient ossification of the anterior frontanelle is major features. Individuals affected by this disease have a characteristic frontal bossing due to bulky forehead, hypertelorism and midfacial hypoplasia [1]. General health is usually good and with normal IQ hence there are usually no severe or debilitating general health effects and hence there is no associated impairment in cognitive or intellectual functioning of affected persons.

Cleidocranial Dysplasia hence in general has dento – facial features including multiple supernumerary teeth, over retained deciduous teeth thereby causing malocclusion or crowding while in some cases due to over retained deciduous teeth and uninterrupted permanent teeth it may be seen as spacing between them. Hence dental management plays as important role in affected persons.

Cleidocranial dysplasia is an inherited autosomal dominant trait, with generation-to-generation transmission. Numerous members of an extended family and a founder effect were initially documented by Jackson in 1951[3].

History of CCD

It dates back to prehistorical times, with first case documented by Greig in 1933[4]. He was a Scottish surgeon who became curator of the Museum of Royal College of surgeons in Edinburgh. Another objective case from ancient Greece is represented by a skeleton of women who lived in Pylos region. Her absent clavicles and stunted stature were thought to be suggestive of CCD [5]. In another reported case of a male skeleton that died due to tuberculosis in 1809 and presently displayed in the Museum of Pathological Academy in Vienna [6, 7], shows manifestations of CCD. However, Meckel's case report of 1760[8], is recognized as the earliest published medical literature. Johann Frederick Meckel the Elder was a professor of anatomy and surgical obstetrics at the University of Halle. Five years after Meckel's article Martin [9] in the year 1765 documented "Natural displacement of the clavicle" in the French literature. There work was more on to non-cranial findings while Scheuthauer [10] in the year 1871 published a combination of clavicular and cranial defects. The Parisian physicians Marie and Sainton[11] documented an affected father and son and titled their article as "On hereditary cleidocranial dysostosis", thereby formally naming the disorder[12]. Extensive minor skeletal involvement was emphasized by Jensen and the name was changed to Cleidocranial Dysplasia [13]. In the year 1995 Mudlos S and coworkers [14] published an article after genetic mapping, in which they established a determinant gene to be chromosomal locus 6p21 [15]. The gene termed RUNX2 (runt – related transcription factor 2) has been sequenced and considerable intragenic heterogeneity has been recognized. It has been shown that the gene product is involved in the control of osteoblastic differentiation and chondrocyte mutation during endochondral ossification [16].

CASE REPORT

A 17 year old male patient reported to our hospital with a complain of small sized teeth since they erupted and preferred to get an orthodontic tooth correction. History revealed a normal eruption of his deciduous teeth while none of the teeth has exfoliated but there is spacing between the teeth. The patient is not aware of any permanent erupted teeth but his teeth are yellowish in color and posterior teeth appear to be black suggesting caries. There are no deleterious habits reported by the patient. He is responsive, agile, while appears to be short statured among his ethnic group. Certain prominent features noticed was frontal bossing with a prominent depression in the midline of forehead. Due to frontal bossing he appears to have sunken eyes, broad nose, concave facial profile and brachycephalic head as seen in figure 1. General examination shows short stature, short neck, and drooping shoulders which are evident from figure 2, while there are bulged extremities of hand and feet as noticed in figure 3 and 4. Intra oral examination shows shallow maxillary arch, generalized spacing between teeth, generalized teeth mobility and only permanent first molars erupted as seen in figure 5 and 6. Patient had a road traffic accident 4 years ago and his second finger of left hand had to be amputated. On further questioning patient revealed his ability to bring his shoulders abnormally front and almost touching to each other which is expressed in the figure 7. Patient secondary growth appears adequate and questioning him found no abnormality. Patient was advised panoramic radiograph and few extra oral radiographs like lateral cephalogram, Posterio – anterior and chest radiograph which revealed presence of 16 impacted teeth in the panoramic radiograph as seen in figure 8. His lateral cephalogram as in figure 9 shows presence of radiolucent area in the occipital region posterior to parieto – occipital suture interspersed with highly reticular pattern suggesting Wormian boned. Parietal area appears prominent, frontal sinus is large while mid facial area appears deficient and hypoplastic. Maxillary and mandibular dentition shows mixed erupted, unerupted and impacted teeth with very prominent mental area. Gonial angle is wide and there are open sutures in the fronto – parietal and parieto – occipital region. Posterio – anterior view in figure 10 shows radiolucent reticular thread like areas behind the fronto – parietal sutures suggesting Wormian bone. Radiolucent diffuse, inverted pear shape area is seen at junction of coronal andf lambdoid region. Wormian bone pattern seen on either side of inter parietal suture lower third of face appears longer hence giving an inverted bulb shape outline. The figure 11 shows chest radiograph which shows hypodeveloped clavicles coinciding with his ability to bring his shoulders front while the other skeletal growth appears to be normal.

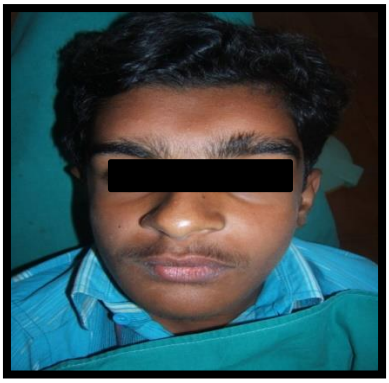

Fig-1: Facial Appearance

Fig-2: Short height

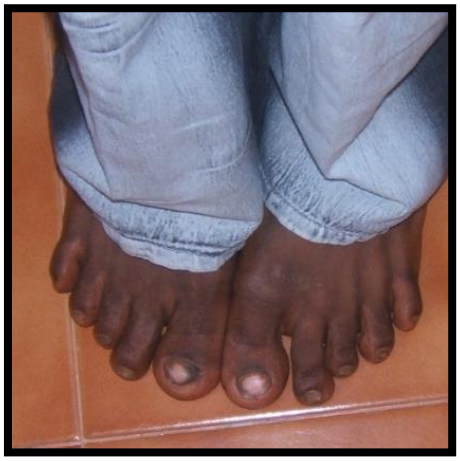

Fig-3: Bulged feet fingers

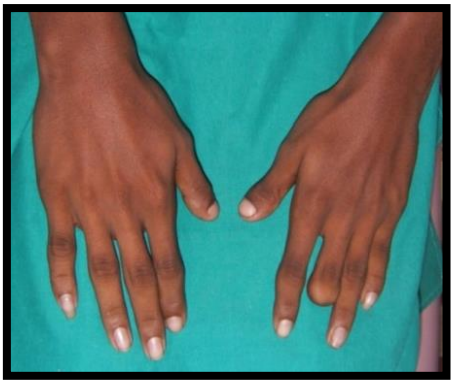

Fig-4: Bulged Hand fingers

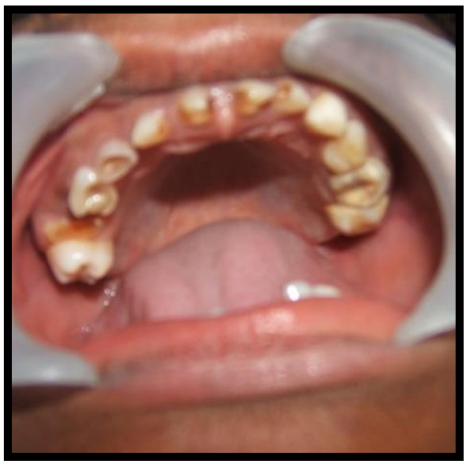

Fig-5: Maxillary Arch

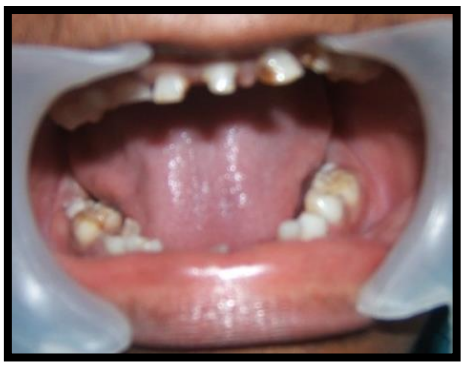

Fig-6: Mandibular Arch

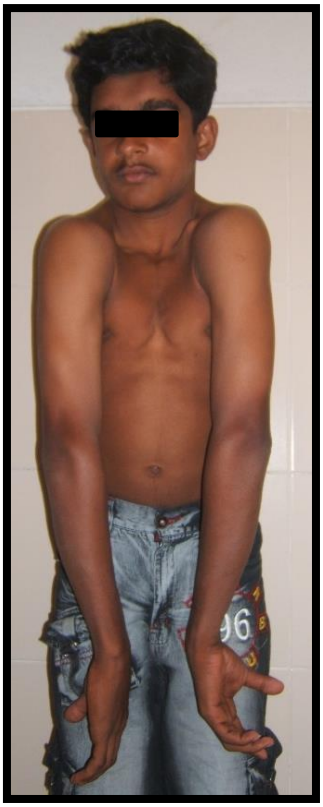

Fig-7: Ability to bring shoulders front

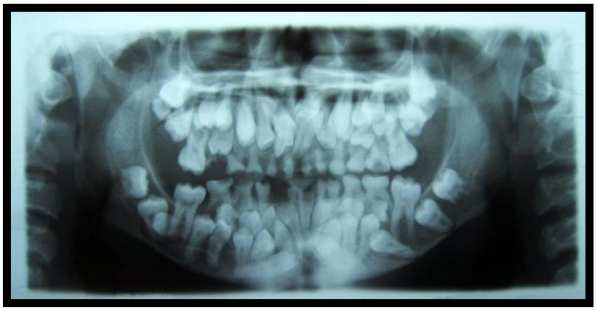

Fig-8: Panoramic radiograph showing multiple impacted and un erupted teeth

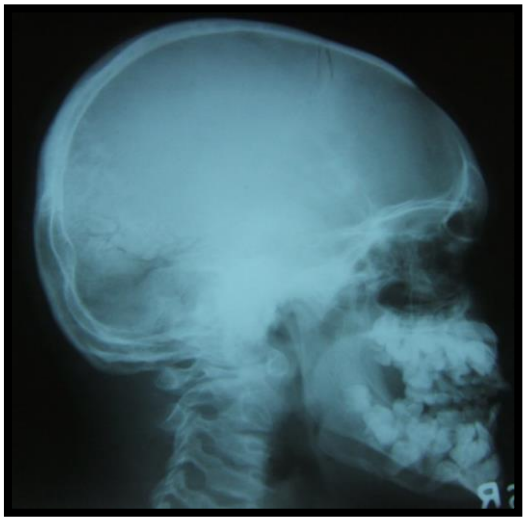

Fig-9: Lateral Cephalogram showing woven bones and deficient mid facial growth

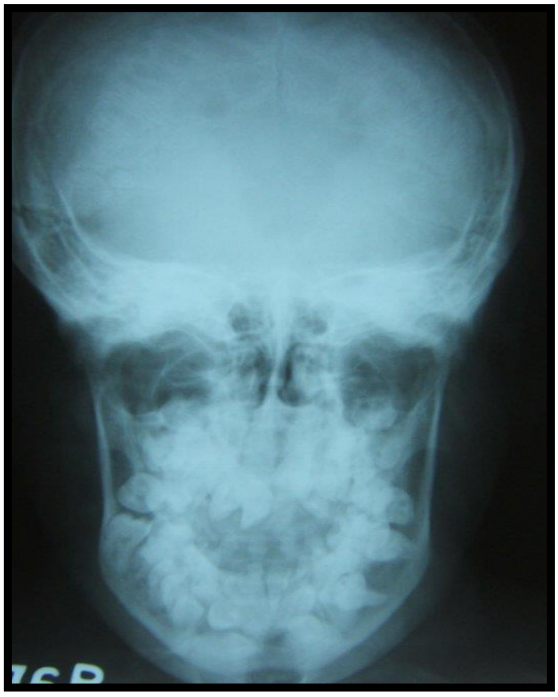

Fig-10: Posterio – Anterior Radiograph showing bulb shaped outline and woven bone

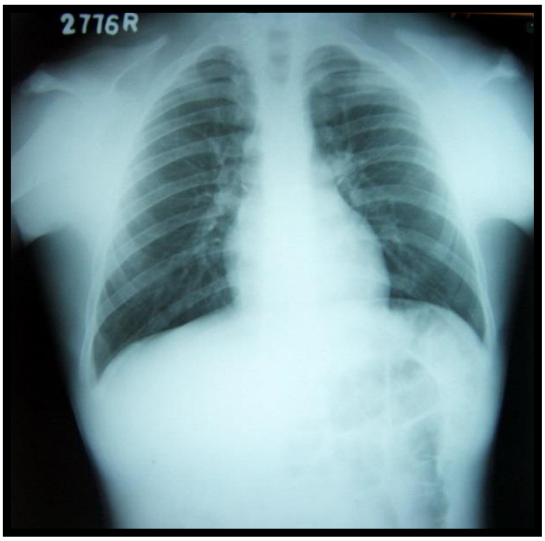

Fig-11: Chest radiograph showing hypoplastic clavicles bilaterally

DISCUSSION

A familial incidence was recorded in approximately 2/3 rd of the reported cases of CCD and the condition was found in as many as five successive generations. When inherited, it appears as an autosomal dominant disease[17,18] In those cases which appeared to have developed sporadically, as with the case presented here, it has been suggested that they represent a recessively inherited disease or more likely either an incomplete penetrance in a genetic trait with variable gene expression or a true new dominant mutation[19]. In our case, the patient did not report

the existence of direct ancestors or descendants who presented any clinical characteristic of CCD.

CCD has been described by many authors and over a period of time the changes have been well documented. The facial features have been similar to our patient as reported in the literature [17, 20, 21]. There is a strong mention of large open frontanelles with prominent forehead and decreased mid facial growth as was seen in our case. Added to this there are reports of frontal depression due to improper or incomplete closure of frontal suture. Most of the literature has cases reported in the early childhood [20, 21] while our case was of a 17 year old patient. There are different instances of patient ability to bring his shoulders forward and since our patient was old he was not able to join the shoulders but was able to bring it abnormally forward and this can be due to his increase age and fusion of other chest bones as evident from the chest radiograph. These patients have normal IQ as was seen in our patient and the secondary growth characters were normal as it is evident from his growth of mustache, hairs on other parts of body apart from a detailed history given by the patient of normal growth in secondary sexual characteristics. There is a generalized failure midfacial growth as seen in our case leading to patent fontanella, metopic suture, wormian bones, nasal deformity [17, 20, 21]. Apart from these there are reports of certain changes like non-union of mandibular symphysis, high arched palate, cleft palate, spina bifida and delayed closure of pubic symphysis which was not seen in our case and this can be due to his increased age which allowed the closure of most of the sutures making the bone remain Wormian in close to suture areas [22]. Final height is significantly reduced in patients with CCD as in our patient the overall height was 153cms. Furthermore there are reports of multiple unerupted and impacted teeth as was seen in our case which had 16 unerupted teeth in his upper and lower jaw and almost equal in all the quadrants. Common complications of CCD include pes planus, genu valgum, shoulder and hip dislocation, recurrent sinusitis, upper airway complications, recurrent ear infection, hearing loss, dental caries, osteomyelitis of the mandible or maxilla, respiratory distress in early infancy etc[23-25]. Even with these complications the life span in such patients is normal. None of the above mentioned complications were found in the present case.

CONCLUSION

CCD is a relatively rare clinical finding and often diagnosed after a confirmed radiographic evaluation. The clinician should be aware of the characteristic features of CCD for early diagnosis and initiating the appropriate treatment approach. It should be considered in the differential diagnosis of short stature with skeletal abnormalities like large fontanella and wormian bones. Early diagnosis allows a proper orientation for the treatment, offering a better compliance to the patient and with anticipatory guidance; people with CCD lead healthy and productive life.

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