

CLEIDOCRANIAL DYSPLASIA- LITERATURE REVIEW AND AN EVIDENCE BASED CLINICAL REPORT

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ABSTRACT

Cleidocranial dysplasia is an autosomal dominant disorder affecting the craniofacial region and clavicles along with other skeletal abnormalities. The Dental surgeon, is usually the first professional whom patients look for to solve the problem, since there is a delay in the eruption or an absence of permanent teeth. This peculiar case has been reported along with literature review to focus on the unusual findings so that the dental professionals become familiar with such features on confronting subjects with Cleidocranial Dysplasia.

Keywords: *Cleidocranial Dysplasia, Aplastic clavicles, Multiple missing teeth, Multiple impacted teeth.*

INTRODUCTION

Cleidocranial dysplasia (CCD) is a rare developmental defect of autosomal dominant inheritance¹ and affects 1 per million individuals worldwide² with no predilection for gender or ethnic group.³ CCD is a bone disorder caused by a defect in the CBF1 gene of chromosome 6p21 which controls osteoblastic differentiation and appropriate bone formation.⁴ This rare bone disorder was first reported in 1760 by Meckel and was later reported by Martin in 1765.⁵ The descriptive term Cleidocranial dysostosis was coined by Marie and Sainton.⁶ Kallialla et al⁷ suggested the genetic mutation as an etiological factor of the disease. In 1946 Forlan⁸ reported that it is a genetic disease with an autosomal dominant inheritance. Later on it was found that there is generalized dysplasia of the bone in this condition and therefore the term "Dysostosis" was replaced with "Dysplasia".⁹ Hesse was first to describe in detail the defects of dentition and jaws associated with cleidocranial dysplasia.¹⁰

CCD individuals are usually 3-6 inches shorter than family members but the most characteristic and pathognomonic feature of this disorder is hypoplasia or aplasia of the clavicles, which results in hypermobility of the shoulders allowing the patients to approximate the shoulders in the midline.¹¹

Muscle attachments to the clavicles may also be dysplastic, leading to distortion of the neck. Defects

of the cervical and lumbar vertebrae are included in the clinical findings. Absence of the pubic symphysis and hypoplasia of the pelvis is common in females. Frequently, flat feet (57%), knock knee deformity (28%) and scoliosis (18%) are found in children younger than 5 years of age. Delayed ossification of the cranial sutures and fontanelles occurs, and may remain open throughout life. Characteristic facial features may include a wide, short skull (brachycephaly); a prominent forehead; wide-set eyes (hypertelorism); a flat nose; and a small upper jaw.¹²

The face appears small in relation to the cranium with hypoplastic maxillary, lachrymal, nasal, and zygomatic bones. Craniofacial growth is affected with the patient displaying, frontal and parietal bossing, relative prognathism due to short, underdeveloped maxilla and depressed nasal bridge.¹¹ The maxillary sinuses may be small or missing. The palate may be abnormally high, and, occasionally, a cleft palate has been reported.

About 94% persons with CCD have dental abnormalities which include delayed loss of the primary teeth; delayed appearance of the secondary teeth; unusually shaped, peg-like teeth, malocclusion and supernumerary teeth,¹³ sometimes accompanied by follicular cysts.^{3,14}

In addition to skeletal and dental abnormalities, people with cleidocranial dysplasia may have hearing loss and be prone to sinus and ear infections.¹⁵ Some young children with this condition are mildly delayed in the development of motor skills such as crawling and walking, but intelligence is unaffected with overall good prognosis and normal life expectancy.^{16,17}

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The differential diagnosis of CCD include Crane-Heise syndrome , Mandibuloacral dysplasia, Pycnodysostosis, Yunis Varon syndrome, CDAGS syndrome, Hypophosphatasia, Parietal foramina with cleidocranial dysplasia (PFMCCD).¹⁸

CCD can be diagnosed by examination of the cranium, face and clavicles including shoulder mobility. Intra oral examination will help to compare the eruption status of the dentition and the patient's chronological age. However the most important and reliable means to confirm diagnosis is the radiographic evaluation of patients, since radiological findings of cleidocranial dysplasia are pathognomonic, i.e. broad sutures, large fontanels persisting into adulthood, numerous wormian bones and numerous unerupted supernumerary teeth.^{7,19}

Treatment of the oral manifestation depends on the services of an integrated team, made up of a pedodontist, orthodontist and oral and maxillofacial surgeon.²⁰

Early referral allows for timely planning of necessary procedures. The dental problems that need to be addressed include the retention of deciduous dentition, the presence of supernumerary teeth, and the non-eruption of the permanent dentition. The goal of treatment is to improve appearance and to provide a functioning masticatory mechanism. The goals may be achieved with prosthetic replacements, with or without prior extractions; by removal of the supernumerary teeth followed by surgical repositioning of the permanent teeth; and by a combination of surgical and orthodontic measures for actively erupting and aligning the impacted permanent teeth.^{21,22}

Sinus and middle ear infections need aggressive and timely treatment; tympanostomy tubes should be considered when middle ear infections are recurrent.²³

CASE REPORT

A 27 years young man reported to the Department of Oral and Maxillofacial Surgery, Khyber College of Dentistry, Peshawar on 15-9-2010. His chief complaint was painful left upper jaw and he desired to remove one of his teeth in the same region. He gave a history of extractions of few teeth 3 months back from upper left jaw but could not recall the number of teeth extracted. Upon general examination, he was noted to have short stature. The skull was brachycephalic. The midface was depressed because

of underdeveloped maxillae, so the mandible appeared prognathic (Figure 1). He had hypermobility of the shoulders allowing the patients to approximate the shoulders in the midline. The other extraoral findings were long neck, frontal bossing, broad nasal bridge and hypertelorism (Figure 2).

His intra oral examination revealed poor oral hygiene, multiple missing teeth and a grossly carious 26 (Figure 3). His orthopantomogram outlined the presence of total 39 teeth in both the jaws. Out of these, 24 were impacted and 15 were seen in the oral cavity. His OPG also confirmed that few teeth were extracted from upper left premolar region but the exact number could not be ascertained. Four follicular cysts were also noted. The gonial angle appeared extremely rounded (Figure 4). In lateral skull radiograph, there was depression in anterior fontanelle region. The hypoplastic appearance of maxilla was also very clearly shown on the lateral skull radiograph (Figure 5) while his P.A. skull radiograph also showed open anterior fontanelle and sutures of skull (Figure 6). Chest radiograph of the patient showed that both his clavicles were absent (Figure 7). All his clinical features and investigations matched the classical description of CCD.

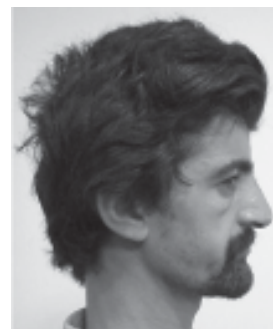


Fig. 1: Patient's profile showing concavity due to underdeveloped maxillae.



Fig. 2: Patient showing hypermobility of both shoulders.

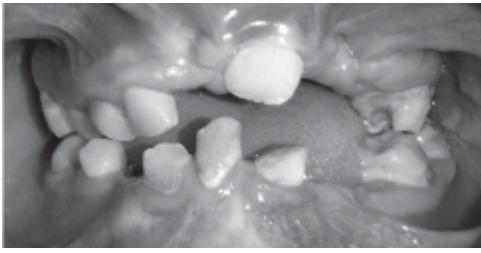


Fig. 3: Intra oral photograph showing multiple missing teeth, poor oral hygiene and grossly carious 26



Fig. 4: Orthopantomogram showing multiple impacted teeth along with Supernumerary teeth and rounded gonial angles



Fig. 5: Lateral skull view showing depression in anterior fontanelle and underdeveloped maxilla



Fig. 6: P.A. skull radiograph showing open anterior fontanelle and sutures of skull.



Fig. 7: PA Chest View showing missing clavicles.

The patient was medicated for his acute pain and an elaborate plan was made to treat the patient accordingly including his oral rehabilitation.

DISCUSSION

CCD is rare autosomal dominant condition and so far over 1000 cases have been reported in literature worldwide.⁴ Only few were reported in India²⁴ but in Pakistan the first case was reported in 2005.²⁵ The rare occurrence of this particular disease prompted the author to report this case.

The author observations in this case were multiple missing teeth and at the same time large number of impacted teeth in both maxilla and mandible. Similar findings were reported by Richardson²⁶ and Frame.²⁷ Most of the supernumerary teeth resembled the premolar teeth. These unerupted teeth interfered with day to day dental care of the adult patient. The total number of teeth were 39 which is not a very high figure when compared with 64 teeth reported by Rizvi et al.²⁸ High number of teeth was also reported by Frame,²⁷ Richardson³⁰ and Jensen.³⁰

Although the patient was in his late twenties, his fontanelles were still open. This feature is also reported in previous recorded cases.^{31,32} Along with this, there were four follicular cysts and bilateral missing gonial angles. In this case, the clavicles were absent which allowed hyper-mobility of both the shoulder. However, cases have been reported where the clavicles were hypoplastic rather than aplastic with resultant hypermobility to a lesser extent.^{27,32}

The present case initially refused to undergo surgery for treatment of follicular cysts but later on agreed after counseling. He is primed for enucleation of four follicular cysts and extraction of all involved teeth followed by fabrication of prosthesis to replace the missing dentition. Patients with CCD if diagnosed at an early mixed dentition stage, will help to provide surgical/orthodontic treatment at pro-per time before adulthood. Long term follow up of CCD patients is

also essential to check the development of follicular cysts.

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