CASE REPORT

Cleidocranial dysplasia: A rare case report

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Introduction

Cleidocranial dysplasia (CCD) is an infrequent inherited anomaly having characteristic features as multiple skeletal defects, delayed closure of the fontanelle, the presence of open skull sutures and multiple wormian bones. As a prominent feature of this disorder frontal bossing, brachycephaly and hypertelorism occurs due to delayed closure of anterior fontanelle, metopic sutures, and reduced growth of dysplastic skull base. Thoracic cage is found to be small, bell-shaped with short ribs and the most prominent feature is hypermobility of shoulder girdle which occurs due to incomplete or complete absence of clavicles bone. Apart from clavicle, the other bones involved are long bones, vertebral column, pelvis, and the bones of hands and feet. Intraorally mixed dentition was appreciated due to persistent retention of deciduous dentition and delayed eruption of permanent teeth as a result of which it shows a large number of unerupted supernumerary teeth.

Case Report

A male patient with 35 years of age stated to the outpatient department of our department with missing teeth in his upper and lower jaws as his chief complaint and wants the fabrication for the same. On general examination, he was of short height with normal gait with hypermobility of the shoulders. On extraoral examination, appearance was found to be bossing of the frontal bone, hypertelorism, and a wide nasal bridge with little depression and due to the underdeveloped maxilla, mandible appears to be prognathic. Intraorally teeth present were as follows: 51, 52, 53, 55, 14, 16, 17, 61, 63, 26, 27, 75, 31, 34, 36, 83, 85, 41, 44, and 46 which revealed that the maxillary arch had few lost teeth and partially erupted premolars. Other clinical features include narrow and deep palatal arch. Due to poor oral hygiene, the residual teeth were affected with caries and generalized periodontitis. On the basis of the clinical examination, the patient was provisionally diagnosed as CCD and its differential diagnosis includes hypohidrotic ectodermal dysplasia, focal dermal hypoplasia, Apert syndrome, and mandibulofacial dysplasia for the same.

On examination, orthopantogram revealed clusters of impacted supernumerary teeth in the edentulous areas, mostly resembling premolars which is considered to be one of the striking features of the syndrome, CCD.

Abstract

Cleidocranial dysplasia (CCD) is a congenital incongruity which involves basically skeleton and teeth. CCD is an infrequent seen autosomal dominant syndrome with frequency ratio of 1:1,000,000 individual and occurs due to alteration in a gene on 6p21 coding transcription factor CBFA1. The most striking features comprises orofacial expressions including enamel hypoplasia, retained deciduous teeth, and impacted permanent and supernumerary teeth, and hypoplasia or aplasia of clavicle bones with the consequential capability of the patient to move his shoulders toward each other. This case has been dealt with a male patient with an age of 35 years who stated to our department with a missing upper and lower teeth as his chief complaint. CCD is an infrequent genetically involved disorder which affects both genders equally and its earlier diagnosis is important for initiating the suitable treatment.

Keywords:
Clavicle bones, cleidocranial dysplasia, impacted, supernumerary teeth

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On comparing the clinical and radiological findings, the patient was diagnosed as CCD. The patient was referred to the departments of periodontics, oral and maxillofacial surgery, and prosthodontics, where oral prophylaxis, extraction of the mobile and retained deciduous teeth was performed which was followed by insertion of a prosthesis [Figure 6].

**Discussion**

CCD (Cleido = collar bone + cranial = head + dysplasia = abnormal forming) is an infrequent disorder of bone with an autosomal dominant inheritance. In 1765, this disorder was
First described by Martin on patient with inborn absence of clavicle.\(^6\) The term cleidocranial dysostosis was coined by Marie and Sainton in 1898.\(^5\) The term dysostosis and dysplasia has been derived from a Greek word osteon means defective ossification of fetal cartilages and plassein means abnormal growth or development, respectively; at molecular level, it means alteration in size, shape, and organization of adult cells.\(^6\) Its etiology is unknown but could be due to a mutation of the RUNX2 (core binding factor alpha-1) gene located at chromosome 6p21. This gene encodes a protein, which normally guides osteoblastic differentiation and appropriate bone formation.\(^8\) In CCD, there is no gender predilection. The significant clinical features include moderately short stature, open fontanelle/delayed closure, sutures remain open, wormial bones, brachycephaly (shape of the skull shorter than normal) and a frontal bossing (protruded forehead), hypoplasia of maxillae, lacrimal, nasal, and zygomatic bones, underdeveloped and narrow paranasal sinuses, ocular hypertelorism and a mild exophthalmos, imperfections in vertebral column, pelvis, and long bones, as well as bones of the digits and may or may not be associated with mental retardation but most patients have normal intelligence.\(^1\) But in the patient reported no abnormality detected in a vertebral column, long bones and digits and the presence of wormian bones and an open anterior fontanelle.

Intraorally, the most characteristic feature is multiple impacted supernumerary teeth and others include over retained deciduous teeth without any resorption in roots, delayed/retarded eruption of permanent teeth, crypt formation around impacted and ectopic teeth, high narrow arched palate and infrequently cleft palate has been reported, partial anodontia, skeletal Class III tendency due to the underdeveloped maxilla, delay in the root development of permanent teeth and the roots are short and thinner than the usual and may be deformed, absence or lack of cellular cementum on the roots of the permanent teeth with no increased thickening of primary acellular cementum.\(^11\) The case reported had a total of 58 teeth in both jaws, with 38 impacted teeth and 20 erupted teeth.

The differential diagnosis of CCD includes Crane-Heise syndrome, mandibulolacral dysplasia, pycnodysostosis, Yunis-Varon syndrome, CDAGS syndrome, and hypophosphatasia.\(^12\) All of these are autosomal recessive disorders with some peculiar features that makes it distinguishable from CCD as Apert syndrome characterized by craniosynostosis, craniofacial abnormalities, and symmetrical syndactyly of the hands and feet; pycnodysostosis mainly including a short limbed stature, acro-osteolysis, osteosclerosis, and bone fragility; and craniofacial dysostosis, which is characterized mainly by premature craniosynostosis, with other abnormalities.\(^13\)\(^13\)

Its treatment is a multidisciplinary approach which includes the equal contribution of periodontists, oral and maxillofacial surgeons, orthodontists, and prosthodontists. The planning of CCD varies from needs of the patient, age at which is diagnosed, and social and economic circumstances. Although there are various treatment modalities, but primarily it is concerned with the craniofacial restoration with dental function along with aesthetics.\(^14\) The treatment procedure involves restoration of the curious deciduous teeth, as their extraction does not necessarily induce the eruption of the permanent teeth orthodontic treatment is usually indicated to direct the eruption of the malposed and often impacted teeth. It also involves the orthognathic surgery to fix the maxillary hypoplasia and extraction of some of the supernumerary teeth may be needed. Surgical removal of impacted teeth may be done in association with orthodontic and/or prosthetic therapy. Timely intervention is critical. Dentures can be fabricated over unerupted teeth. In this case, all carious teeth were restored, all mobile and deciduous teeth that were not in occlusion were extracted, and a removable prosthesis was given.

**Conclusion**

CCD, a rare disorder is largely diagnosed incidentally. But for rendering orthognathic and orthodontic correction, such patients should be analyzed at an early mixed dentition stage as age of the patient plays an important role in the treatment. The correlation of patient’s family history, excessive mobility of shoulders and radiographic pathognomonic findings of the chest, skull, and jaws are useful for the confirmation of the diagnosis.

**References**
