

CLEIDOCRANIAL DYSPLASIA- A CASE REPORT AND LITERATURE REVIEW

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ABSTRACT

Cleidocranial dysplasia is a rare congenital skeletal disorder, associated with hypoplasia or aplasia of clavicles, delayed closure of cranial sutures and fontanelles, frontoparietal bossing, delayed exfoliation of primary dentition, delayed or failure of eruption of permanent teeth, and presence of multiple supernumerary teeth. The disorder is caused by mutation in the CBFA1 gene, on the short arm of chromosome 6p21. Estimated prevalence of cleidocranial dysplasia is one per million, without gender or ethnic predilection. A multidisciplinary approach is often required for dental management of multiple supernumerary teeth. We report clinical and radiographic presentation and surgical management of multiple supernumerary teeth in an 11-year-old child with cleidocranial dysplasia.

Key Words: *Cleidocranial Dysplasia, Cleidocranial Dysostosis, Supernumerary, Teeth, Mandible, Jaws.*

INTRODUCTION

Cleidocranial dysplasia (CCD) is a rare skeletal disorder caused by a defect in the CBFA1 gene on chromosome 6p21. This gene controls osteoblastic differentiation, appropriate osteogenesis and odontogenesis via participation in odontoblast differentiation, enamel organ formation and dental lamina proliferation. CCD has an estimated prevalence of 1:1000000. CCD is characterized by short stature, large head with frontal and parietal bossing, delayed closure of cranial sutures and fontanelles, hypertelorism with depressed infraorbital area, broad based nose with depressed nasal bridge and aplasia or varying degrees of hypoplasia of clavicles.¹

The orodental manifestations are a narrow, high arched palate, prolonged retention of primary teeth, delayed or failing eruption of permanent teeth, presence of numerous unerupted permanent and supernumerary teeth.^{1,2} Various theories have tried to explain the devel-

opment of supernumerary teeth; such as Phylogenetic reversion theory, Dichotomy theory and dental lamina hyperactivity theory.^{3,4} Failure of permanent tooth eruption in patients with CCD has been attributed to insufficient bone resorption.²

Dental treatment options for a patient of CCD include full mouth extractions with fabrication of denture, auto transplantation of selected impacted teeth with/without prosthetic restoration and removal of primary and supernumerary teeth followed by orthodontically assisted eruption of impacted permanent teeth.⁵ We report clinical presentation and surgical management of multiple supernumerary teeth in both jaws of an 11-year-old child with CCD.

CASE REPORT

An 11-year-old female patient, reported to Military Dental Centre Gujranwala, Pakistan, in January 2016; with complaint of failure of eruption of permanent dentition. A thorough medical and dental history was recorded along with clinical and radiological examination.

Our patient was given birth as a full term baby by her 34-year-old mother through an uneventful caesarian section in a military hospital. Parents of the patient did not know about her birth weight. She had three sisters; one elder and two younger to her. Her elder sister also had delayed exfoliation of her primary teeth and delayed eruption of permanent successors.

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The patient was aware of her ability to approximate her shoulders in the midline anteriorly. She was an intelligent student and used to pass in exams with distinction. On clinical examination, she was of average built and height with normal facial profile and competent lips. She had frontal bossing, telecanthus, slightly depressed nasal bridge and prominent nasal tip. Her head circumference, body weight and height were 54cm, 36 kg and 134cm respectively. Her mouth opening was normal and TMJ examination was insignificant. There was no cervicofacial lymphadenopathy. On palpation, both the clavicles were found missing. Skin elasticity, joint mobility and finger count were normal. Her intelligence was subjectively normal. Rest of the general physical examination was unremarkable.

During intraoral examination, the patient was found to be in mixed dentition stage with following teeth fully erupted.

6,e,d,c,b,a	a,b,c,d,e,6
6,e,d,c,b,1	1,b,c,d,e,6

She had Angle's class I occlusion with edge to edge incisors and bilateral posterior cross bite. On radiographic examination, Orthopantomograph of the patient revealed unerupted permanent teeth and 12 supernumerary teeth, distributed in both jaws; 9 in mandible and 3 in maxilla (Fig 1). Analysis of Lateral cephalometric radiograph revealed a hypoplastic maxilla. Chest radiograph (posteroanterior view) confirmed bilateral absence of clavicles (Fig 2). A cone beam computed tomography scan of the patient was recorded which revealed that all supernumerary teeth were located lingually in both jaws.

After discussion with Orthodontist, it was decided to remove all the supernumerary teeth in the mandible and maxilla, leaving all the primary teeth undisturbed with exception of upper and lower primary incisors. It was decided to wait for a reasonable period of time for spontaneous eruption of permanent teeth. Meanwhile maxillary expansion will be carried out with a suitable orthodontic appliance (palatal expansion plate/Hyrax appliance). Patient was prepared to be operated under general anaesthesia after informed written consent. Treatment was carried out as per preoperative planning (Fig 3). Impacted supernumerary teeth were approached through both buccal and lingual cortical plates after removal of cortical bone using round carbide bur under copious irrigation with normal saline. Post operative recovery was uneventful. The patient will be followed at regular intervals for spontaneous eruption of permanent dentition and if needed, their eruption will be assisted by orthodontic intervention.



Fig 1: Orthopantomogram showing unerupted permanent and supernumerary teeth



Fig 2: Clinical and radiographic evidence of clavicular absence



Fig 3: Post operative Orthopantomogram

DISCUSSION

CCD is a rare skeletal disease caused by mutation in the *CBFA1* gene on chromosome 6p21, affecting both sexes and all ethnic groups equally with a prevalence of approximately 1 in 1 million individuals. In Pakistan, the first case of CCD was reported in 2005.⁶ The rarity of CCD prompted the authors to report this case. It presents with characteristic skeletal defects of several bones, primarily affecting bones that undergo intra-membranous ossification such as clavicles, cranium, face and pelvis.⁷

Diagnosis of CCD in our patient was made on clinical appearance and confirmed radiologically. Many of the characteristic features were present in our patient

such as frontal bossing, hypotelorism, depressed nasal bridge, delayed eruption of permanent teeth, presence of multiple supernumerary teeth and ability to approximate shoulders in front of the chest.

There was positive family history of delayed eruption of permanent teeth of the elder sister of our patient. Height, weight and head circumference of the patient were within normal limits. Lack of abnormal skin elasticity and hypermobile joints ruled out the possibility of Ehlers-Danlos syndrome clinically which may be associated with multiple supernumerary teeth.⁸ Other syndromes associated with multiple supernumerary teeth are Gardner's syndrome, Down syndrome and Noonan's syndrome.⁹ Both clavicles were absent in our patient which allowed approximation of shoulders in front of chest. However, cases have been reported in literature where the clavicles were hypoplastic rather than aplastic with resultant hypermobility to a lesser extent.¹⁰

The most accepted theory to explain the development of multiple supernumerary teeth is the 'dental lamina hyper activity theory' which states that lingual extension and proliferation of dental lamina gives rise to supernumerary teeth.² In case of our patient all the supernumerary teeth were positioned lingual to other teeth in both jaws which is in agreement with the above mentioned theory. The reason for failure of tooth eruption in patients of CCD is not well understood however insufficient alveolar bone resorption has been proposed as a contributory factor.⁵

Owing to the rarity of the condition, guidelines for treatment for clavicular, skull and other bone anomalies associated with CCD does not exist in the literature.¹¹ However treatment options for correction of dental abnormalities include full mouth dental extractions with fabrication of denture, auto transplantation of selected impacted teeth with/without prosthetic restoration and removal of primary and supernumerary teeth followed by orthodontically assisted eruption of impacted permanent teeth. The latter option seems to be the treatment of choice.^{5,12}

We surgically removed all the 12 supernumerary teeth under general anaesthesia, to remove the hindrances and facilitate spontaneous eruption of permanent successors. For intraoperative identification of impacted supernumerary teeth and their subsequent removal, bur holes were made in both buccal and lingual

cortical plates. We left most of the primary teeth intact in order to avoid long edentulous spans and let the roots of primary teeth act as guide for natural spontaneous eruption of permanent teeth. Maxillary expansion will be carried out with a suitable orthodontic appliance after completion of healing. The patient will be followed for a period of 2 years for spontaneous eruption of permanent successors and in case of delayed or failed eruption; a decision regarding their extraction or orthodontically assisted eruption will be made after consultation with the orthodontist and discussion with the patient.

Early diagnosis of CCD is essential for introduction of appropriate treatment approach based on interdisciplinary cooperation between orthodontists and oral and maxillofacial surgeons. Early treatment, before adulthood can prevent short lower facial height and mandibular prognathism.

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| 1 Sarfaraz Khan: | Main author. |
| 2 Syed Asif Haidar Shah: | Contribution in writing the manuscript. |
| 3 Farman Ali: | Proof reading of manuscript. |
| 4 Dil Rasheed: | Conception of idea. |