Cleidocranial Dysplasia: Report of Two Cases

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ABSTRACT
Cleidocranial dysplasia constitutes a congenital disorder manifested primarily in the development of facial and cranial bones, as well as partial development or complete absence of the clavicles and problems also arise on the number and eruption of teeth. It is a rare disease with autosomal dominant but 40% cases represented spontaneous mutations. This disease has no sex predilection. It is characterized by a generalized skeletal dysplasia.1

In this article we are reporting two cases of cleidocranial dysplasia in son and father with characteristic clinical and radiographic features.

Keywords: Cleidocranial dysplasia, Marie and Saintons disease, Scheuthauer-Marie-Sainton syndrome, Mutational dysostosis, Cranioleiodysostosis.

INTRODUCTION
Cleidocranial dysplasia, a rare disease with autosomal dominant trait, shows presence of hypoplastic and malformed clavicles with narrow thorax allowing the movement of shoulders up to the medial plane of the body without any discomfort.2 The absence of clavicles occurs in 10% of cases. Defects in vertebral column, pelvis and long bones as well as in bones of digits are relatively common. In some cases, muscles are underdeveloped.3

These patients have delayed ossification of skull with excessively large fontanells. The sutures can also remain opened, and the sagittal suture presents itself depressed giving the cranium a flat appearance sometimes referred as “Arnold head”. Skull appears brachycephalic with narrowing of skull in antero-posterior direction and increase in width of skull.4 Frontal, parietal, occipital bones are prominent with ocular hypertelorism and paranasal sinuses are underdeveloped. Calvarial thickening in supraorbital part of frontal bone, squamous part of temporal bone and occipital bone is evident with faulty development of foramen magnum.5

These patients show high arched, narrow and deep palate with high prevalence cleft palate. The maxilla can be underdeveloped and shorter in relation to normally developed mandible resulting in a pseudomandibular prognatism [class3]. The zygomatic and lacrimal bones can also present themselves underdeveloped. Median mandibular suture closes late or not at all. Mandible often shows coarse trabeculation with areas of increased density, narrow ascending rami and slender, pointed coronoid process. Patients also exhibit short lower facial height, acute gonial angle, anterior inclination of mandible, inadequate vertical growth of maxilla and hypoplastic alveolar ridge development.3

It was hypothesized that the dental lamina for both the primary and permanent dentition is normal, but does not resolve completely and therefore may form supernumerary teeth.6 The dental eruption is retarded, with absence of root resorption in the deciduous, also associated with hypodontia and dentigerous cyst. The unerupted teeth are displaced to occupy an oblique or horizontal position in jaws resulting in tilting of erupted teeth. In some cases, crypt formation around impacted teeth is also evident. There is absence or paucity of cellular cementum on roots of permanent teeth, which correlates with failure in eruption of teeth.5,7

CASE REPORT
32-year-old male patient reported to department of oral medicine and radiology at GDC&H Mumbai with chief complaint of multiple erupted teeth and patient wanted restoration and replacement of decayed and missing of teeth. Patient visited dentist few years back and got replacement of lower anterior teeth with fixed prosthesis. Medical history was not significant. Patient was averagely built. Patient has got unusual mobility of the shoulder and was able to bring his shoulder forward until they meet in the midline without discomfort.

Extraoral examination showed brachiocephalic head, nasal bridge was broad and depressed (Figs 1 and 2). Intraoral examination reveals multiple over retained deciduous and multiple permanent teeth along with supernumerary teeth, hypoplasia with 11 and 21, caries with 12, 16 and 37. Fixed prosthesis seen in lower anterior region extendind from 15 to 25 region. Crowding seen in lower anterior region. Oral hygiene was poor. Presence of generalized gingivitis and periodontitis (Fig. 3). Hard palate was high and narrow arch (Fig 4).

Patient’s father also gave history of multiple erupted teeth and was able to bring his shoulder to the midline without discomfort (Fig.5). Patient’s father was complete denture wearer with broad and depressed nasal bridge (Fig. 6). Intraoral examination shows six teeth in upper anterior region. Hard palate was high and narrow arch (Fig.7) and lower jaw was edentulous (Fig. 8).

OPG of the patient showed multiple over retained deciduous teeth and multiple unerupted impacted permanent as well as supernumerary teeth present in both jaws (approximately 45 teeth). Fixed prosthesis was present in the anterior region. Rami were long and narrow (Fig. 9). Patient’s father’s OPG showed multiple impacted permanent teeth in both jaws. Few teeth were horizontally placed and one tooth was inverted (Fig. 10).
Fig. 1: Broad and depressed nasal bridge and approximation of shoulders in front of chest

Fig. 2: Lateral view of face

Fig. 3: Multiple over retained deciduous and multiple permanent teeth along with supernumerary teeth in son

Fig. 4: High arched, narrow and deep palate in son

Fig. 5: Approximation of shoulders in front of chest

Fig. 6: Lateral view of face
DISCUSSION

This pathology was first described in 1765 whereas only in 1898, Marie and Sainton had described cases of the disease and associated them with patterns of inheritance. Later, Bauer apud Kalliala (1962) suggested the genetic mutation as an etiological factor of the disease. In 1946, Lasker apud Forlan (1962) had concluded it was a genetic disease with an autosomal dominant inheritance.8

This syndrome is having familial pattern and transmitted as an autosomal dominant trait. Chromosomal abnormality in long arm of chromosome 8[q22] and long arm of chromosome 6 have been reported to be associated with this syndrome. Three genes were located in area 6p21, these are TCTE1, MUT and Cbfa1 (Run 2) from which the gene for the transcription factor Cbfa1 (core binding factor) was thought to be most likely responsible for the pathogenesis of the disease.

The abnormal process of osteoblastic differentiation was found responsible for the anomalies of bone development and growth. In particular, due to the insufficiency of transcription factor Cbfa1, its target genes are not activated, namely the genes of osteocalcin, VEGF (vascular endothelial growth factor), MMP13 (extracellular matrix metalloproteinase), collagen type 10a1, osteopontin and alkaline phosphatase, which in turn are characteristic cellular products of osteoblastic cells.

The study of the cellular mechanisms of dental eruption on heterozygotic Cbfa1 test animal subjects revealed a decreased number of osteoclasts that contribute to normal resorption of the alveolar bone during tooth eruption. The decreased number of osteoclasts in patients with cleidocranial dysplasia leads to delayed eruption and increased number of impacted teeth.

Cbfa1 regulates morphogenetic mechanisms of growth and development of the skeleton, as well as gene expression in the mesenchymal cells of dental epithelium. Its deficiency leads to manifestation of dental anomalies related to delayed eruption and impaction of the permanent teeth of patients with cleidocranial dysplasia. The presence of supernumerary teeth not only obstructs eruption and produces impaction of permanent successor teeth but also leads to morphological dysplasias of the crowns and more commonly the roots of the permanent teeth due to deficiency of sufficient space for proper development. In conclusion, the deficiency of transcription factor Cbfa1 in cleidocranial dysplasia leads to a deregulation of the morphogenetic mechanisms of skeletal and dental development and growth.1,9

Pycnodysostosis or the Marteaux-Lamy syndrome presents identical features as cleidocranial dysplasia except presence of dwarfism, dense and fragile bones. Mandibuloacral dysplasia has similar presentation as cleidocranial dysplasia except associated with mutations in the genes LMNA or ZMPSTE24 and inheritance is autosomal recessive.6

Planning treatment for a patient with cleidocranial dysplasia is complicated by a host of factors. The treatment plan is largely dependent on both the chronological and dental ages of the patient. The timing of diagnosis is not only important in choosing an appropriate treatment plan but also in attaining a successful result.9 Patient leads a normal life span. Treatment is mainly supportive with no treatment of underlying disorder. Dental treatment requires an interdisciplinary approach involving orthodontist, maxillofacial surgeon and prosthodontist.6
The current “state-of-the-art” treatment involves a combination of orthodontics and maxillofacial surgery. The protocol involves timely extraction of deciduous teeth, staged surgical removal of supernumerary teeth, exposure of selected unerupted permanent teeth and orthodontic forced eruption. Following alignment of all permanent teeth, any underlying skeletal discrepancy (most commonly a Class III skeletal malocclusion) can be corrected through orthognathic surgery after the completion of growth [Lefort I maxillary osteotomy].

Prosthodontic management includes overlay dentures as complete or partial removable dentures that are fabricated over retained teeth or roots that are not specially prepared to accept copings. The complete overlay denture has numerous advantages for the pediatric patient. Enhanced mastication and esthetics are the more obvious benefits and speech also may be improved. The alveolar bone is maintained by the retention of teeth compared to its loss when teeth are extracted. With the retained teeth and their periodontal ligament, there is increased proprioception compared to a complete denture that is fabricated over an edentulous ridge. In some cases, after assessment of bone and performing all necessary investigation, implant is planned onto which implant supported denture is constructed.

Our both cases have similar clinical and radiographic features of cleidocranial dysplasia described in literature. In our cases, we advised oral prophylaxis, restoration of carious and hypoplastic teeth followed by prosthesis.

REFERENCES