Cleidocranial Dysplasia—Unearthing the Treasure

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Abstract

Cleidocranial dysplasia is an autosomal dominant condition caused by mutation of RUNX2, involving the skeleton and the teeth. It presents with a major finding of the hypoplasia or aplasia of clavicles along with late closure of the fontanels, presence of open skull sutures and multiple wormian bones. Although not frequent it can present to the dentist commonly with significant retention of multiple deciduous teeth, impaction or delay in eruption of permanent teeth and often with the presence of supernumerary teeth. Here we report a case of Cleidocranial dysplasia in a 30 year old male.

ABBREVIATIONS

CCD: Cleidocranial Dysplasia

INTRODUCTION

Cleidocranial dysplasia (CCD) is an autosomal dominant condition having a prevalence of less than 1 per million primarily involving bones formed by intramembranous ossification, such as facial bones, cranium and clavicles. CCD is characterized by late closure of the fontanels, persistently open or delayed closure of sutures, hypoplastic or aplastic clavicles, multiple wormian bones and short stature. The vertebral column and appendicular skeleton are also affected, indicating endochondral ossification. Multiple supernumerary teeth are present often accompanied by delayed or disturbed eruption of permanent teeth [1,2].

Cleidocranial dysplasia was first described by Pierre Marie and Paul Sainton in 1898, who coined the term “dysostose cléidocrânienne héréditaire”. It is associated with a spontaneous mutation in the gene coding for osteoblast transcription factor RUNX2. The pathology is due to an early developmental disorder of mesenchyme or connective tissue, causing retarded ossification of bone precursors especially at junctions, which can lead to defective ossification or even failure of ossification of portions of skeletal structures [3-5].

Cleidocranial dysplasia is of clinical significance to dentistry due to the involvement of the facial bones and multiple impacted teeth [6,7]. CCD patients are prone to develop odontogenic cysts from many impacted teeth, so early diagnosis and management is necessary to prevent these complications.

CASE PRESENTATION

A 30 year-old male presented to Department with a complaint of missing teeth in upper and lower jaws. He had few teeth in the jaws since childhood after his milk teeth shed away, which led to an unpleasant smile and psychological trauma while communicating. He insisted for rehabilitation of his missing teeth and smile.

On general examination he had normal built with short stature, short and stout fingers. Facial examination (Figure 1) revealed brachycephalic head, fronto-parietal bossing, prominent orbital ridges, mild hypertelorism and depressed nasal bridge. He also exhibited hypermobility of shoulders, a characteristic sign of CCD due to absence or hypoplasia of clavicles. The oral cavity (Figure 2) had multiple missing permanent teeth in maxillary and mandibular arches with deep palatal vault.

OPG (Figure 3) had multiple unerupted permanent teeth with many impacted supernumerary teeth with loss of gonial angle. Lateral cephalogram (Figure 4) showed underdeveloped paranasal sinuses and zygomaticofacial complex. Open skull sutures, delayed closure of fontanels and multiple wormian bones

Figure 1 Extra oral photograph showing brachycephalic head, fronto-parietal bossing and prominent orbital ridges.
were noted in PA skull view (Figure 5) along with poorly formed paranasal sinuses and zygomatic complex. Chest radiograph showed thin hypoplastic clavicles (Figure 6) with bell shaped rib-cage. Final diagnosis of Cleidocranial dysplasia was made based on clinical and radiological features. Patient was referred to Prosthodontic department for prosthetic rehabilitation.

**DISCUSSION**

Cleidocranial dysplasia (CCD) is a rare congenital defect in which craniofacial growth is affected in many ways. The face usually has a broad forehead with frontal bossing and some degree of hypertelorism. The midfrontal area is poorly developed and shows a frontal groove owing to incomplete ossification of the metopic suture. Closure of the anterior fontanel and sagittal and metopic sutures is delayed often for life. Delayed closure of anterior fontanel and metopic sutures result in frontal bossing. With increasing age wormian bones around the lambdoid suture are seen due to delay in ossification of the skull in the infants. The most striking feature of CCD is the appearance of the brachiocephalic head and face. Narrow abnormal shaped pelvic and pubic bones, small bell shaped thoracic cage with scoliosis have been also reported [5,6,8,9].

In general CCD patients are of a normal intelligence quotient and health. They are slightly dwarfed, slender and have long necks and drooped shoulders. Final height is significantly reduced and usually has a mildly disproportionate short stature with short limbs. Clavicular hypoplasia is one of the hallmarks of CCD. Depending on the degree of clavicular hypoplasia, appearance can range from a dimple in the skin to sloping, almost absent shoulders and the ability to voluntarily bring the shoulders together. Complete absence of the clavicle is rare, whereas hypoplasia of the acromial end is common and bilateral is the rule but not always the case. Hand abnormalities include short, tapered fingers and thumb with shortening of distal phalanges. All other bones of the hands and feet, especially the distal phalanges and the middle phalanges of the second and fifth fingers are unusually short. Clinodactyly of the fifth finger may be present as well [6,8].

The orbital ridges are prominent but the inferior margins
are shrunken into small maxillae, which give the face a flattened appearance resulting in mandibular prognathism. Biparietal cranial enlargement, hypertelorism and absence of the nasal bone are other features in few cases. Frontal and paranasal sinuses are frequently absent or reduced in size [6,8]. Due to facial hypoplasia and underdeveloped paranasal sinuses, upper airway respiratory disorders such as wheezing, sinusitis, chronic throat infection, chronic nasal congestion, sleep apnea, recurrent bronchitis and recurrent pneumonia are commonly seen. Midfacial hypoplasia also results in an increased risk of conduction hearing defect and recurrent ear infection. Pelvic bone deformities may lead to an increased rate of Caesarean section delivery in female adult patients [10].

Intraoral features are characteristic of CCD. The palate is often high arched and clefts of the hard and soft palates have been described. Retention of the deciduous teeth with delayed eruption of the permanent teeth is a relatively constant finding and shows a striking finding of presence of multiple supernumerary teeth, particularly in mandibular premolar and maxillary anterior regions. Morphologically and functionally supernumerary teeth resemble their normal counterparts [6,8,9].

Cleidocranial dysplasia is a systemic disorder that presents itself as a condition in which teeth fail to erupt. The anomaly manifests as almost complete absence of cellular cementum and an increase in the amount of acellular cementum of the roots of the affected teeth. This was thought to be the probable cause for failure of eruption of a significant number of teeth. After the discovery of RUNX2, it is very clear that failure of eruption is mainly due to mutated RUNX2 gene in CCD [5,6,8].

RUNX2 gene is necessary for tooth development which controls the differentiation of precursor cells into osteoblasts, the cells that actually secrete bony matrix and thereby form bone and regulates chondrocyte differentiation during endochondral bone formation. This gene is one of the key mesenchymal factors that influences tooth morphogenesis and subsequent differentiation of ameloblasts and odontoblasts [5,6].

Since radiological findings of CCD are pathognomonic, the radiographic evaluation is the most reliable means to confirm the diagnosis [9]. Radiologically, Clavicular hypoplasia is one of the hallmarks and the combination of normal deciduous teeth, delayed eruption of permanent teeth and multiple impacted supernumerary teeth are diagnostic of CCD. In addition, with the identification of the responsible gene, molecular-genetic analysis has become an indispensable diagnostic tool for early detection of CCD [6,8].

The differential diagnosis of Cleidocranial dysostosis includes Pyknodysostosis, Congenital pseudoarthrosis of the clavicle, Treacher Collins syndrome and Yunis-Varon syndrome [10]. Congenital pseudoarthrosis of the clavicle is probably among the most common conditions to be considered, occurs due to idiopathic failure of coalescence of its ossification centers. In the great majority of cases involvement is unilateral but bilateral version may mimic CCD. Pyknodysostosis is a rare autosomal recessive malformation of osteoclast function having a pathognomonic feature of acro-osteolysis with sclerosis of the terminal phalanges and some features similar to that of CCD. It is characterized by features not seen in CCD: dwarfism, micrognathia, osteosclerosis, hypoplastic or absent distal phalanges and presence of multiple fractures [8,10].

Treacher Collins syndrome is an autosomal disorder with variable expression involving almost exclusively the craniofacial development. It is characterized by antimongoloid slant of the eyes, coloboma of the lid, micrognathia, microtia, ear deformities, hypoplastic zygomatic arches and macrostomia [10]. Yunis-Varon syndrome which is usually a recessively inherited lethal condition is characterized by prenatal growth deficiency, failure to thrive, abnormalities of calvaria, thumbs, toes, clavicle and high neonatal mortality rate. The severity of the syndrome together with limb malformations and a patchy sometimes sclerotic bone structure should make easy distinction from CCD [5,8].

In confirmed cases, genetic counselling for family planning should certainly be advised. It has been proposed that supernumerary teeth should be removed as early as possible because these will always impede normal eruption of permanent teeth. Long term orthodontic and surgical treatment is usually necessary with the aim of actively erupting and aligning the impacted permanent teeth and for patients with compromised esthetics, surgical treatment with orthodontic traction is a convenient and viable alternative [5,8].

CONCLUSION

Being able to diagnose the condition and initiate early treatment is of paramount importance in the preservation of the physical as well as psychosocial well-being of the patient. It is also important to bear in mind that successful treatment in cases such as this can only be achieved through a holistic approach that encompasses all aspects of the condition, including the primary pathology, the deformity itself and as well as the psycho-social implications.

REFERENCES


Cite this article