CASE REPORT

Clinical spectrum of cleidocranial dysplasia

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Abstract

Cleidocranial dysplasia is a rare autosomal dominant condition with generalised dysplasia of bone, characterized by delayed closure of cranial sutures, hypoplastic or aplastic clavicles, short stature, dental abnormalities and a variety of other skeletal abnormalities. We present a 24 year old male patient with classical features of cleidocranial dysplasia.

Introduction

Cleidocranial dysplasia is an autosomal-dominant syndrome of unknown etiology. The inheritance is not fully expressed, as one would expect in a dominant trait, but instead has a variable expressivity. About 35% of cases have no apparent inheritance and probably represent spontaneous mutations. The name of the syndrome is derived from defect of shoulder gridle and rest from the clinical appearance of skull features. This case report is to enlighten the various clinical and radiographic diagnostic parameters which can help in multidisciplinary management.

This disorder of bone formation is mainly characterized by clavicular hypoplasia or agenesis, and frontal bossing. Many patients have residual hypoplastic clavicles rather than complete agenesis, with the residual portion articulating to the sternum. Although a variety of anomalies may be found in other bones, the defect chiefly involves membranous bones, with the skull and clavicles being the chief sites of disorder. The absent clavicles have always been the focus of attention in cleidocranial dysplasia patients. The added motion of the shoulder girdle allows patients to touch their shoulders together in the midline.[1]

Their head will appear brachycephalic with obvious frontal and parietal bossing. The cranium will be enlarged in the anteroposterior dimension and shortened in

the superoinferior dimension, and the nasal bridge will appear flat and broad. There may be a groove in the midline of the forehead, and there may be palpable soft areas in the scalp due to open sutures. The supraorbital and infraorbital ridges are often prominent, and exorbitism may be seen because of a deficient orbital volume due to frontal bone thickening. The characteristic skull appearance is referred to as Arnold head.[1] A variety of additional skull abnormalities also occur and they include calvarial thickening of supraorbital rims, squamous part of temporal bone and occipital bone. The nasolabial angle is usually excessively obtuse. Other rare skeletal abnormalities represent spina bifida, delayed closure of the pubic symphysis, and malformation of the metacarpals and phalanges.

The maxilla will be hypoplastic with a deep, narrow palate that may harbor a submucosal cleft. The anteroposterior deficiency of the maxilla will create a pseudoprognathism. Mandible seems to be enlarged in comparison to maxilla, lacriminal and zygomatic bones are also under developed. Striking oral manifestation will be malocclusion with over retained primary teeth and missing permanent teeth. The delayed exfoliation of the primary dentition may due to delayed root resorption.[2] The multiple unerrupted supernumerary teeth seem to be related to a delayed involution of the dental lamina, which becomes reactivated when the expected permanent tooth develops. Because the dental lamina arises from and forms the teeth from a lingual position, the supernumerary teeth lie lingual and occlusal to the permanent teeth. The multiple unerrupted teeth are believed to be due to absence of cellular cementum and the delayed exfoliation of the primary dentition due to delayed root resorption.[3]

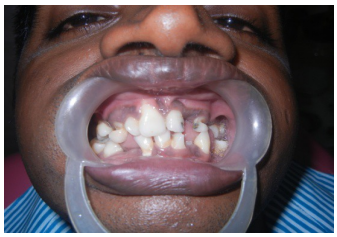
Skull radiographs will show areas of radiolucency corresponding to delayed cranial bone formation. They will also show radiopaque centers of secondary calcification and wormian bones in the sutures. The frontal and sphenoid sinuses will be small or absent, and the mastoid air cells will be missing. The maxillary and ethmoid sinuses will be absent or small.[4]

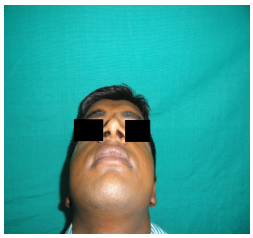
Panoramic radiographs will show, for those in the primary dentition stage (ages 2.5 to 6 years), normal eruption and formation of all 20 primary teeth. Those in the mixed-dentition stage and into adulthood will show numerous unerrupted and supernumerary teeth. The primary dentition will show delayed root resorption and physiologic exfoliation. As a general rule, there is one supernumerary tooth for every expected permanent tooth. This is generally termed as 'double dentition.'

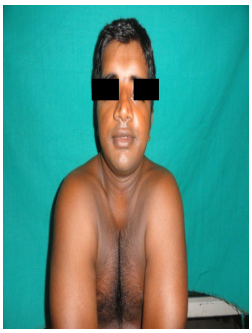
Case report

A 24 year-old male presented to the Government Dental College, Kottayam with a chief complaint of carious and missing teeth in upper and lower jaws. Due to these missing teeth, he had an unpleasant smile that resulted in a psychological trauma to him while communicating with public. Oral cavity examination revealed multiple over-retained deciduous teeth and some missing

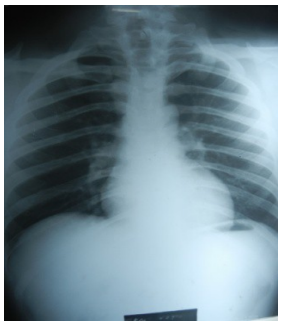
teeth. Facial examination revealed frontal bossing with hypoplastic maxilla, mandible and zygomatic bones. There was exopthalmosis and anti mangaloid slant of eye. Abnormal hypermobility of shoulders, which allowed patient to approximate his shoulders in front of chest, was noticed.

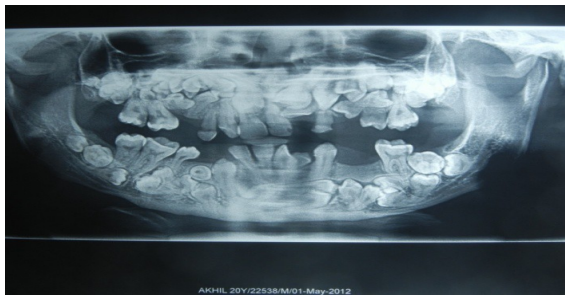






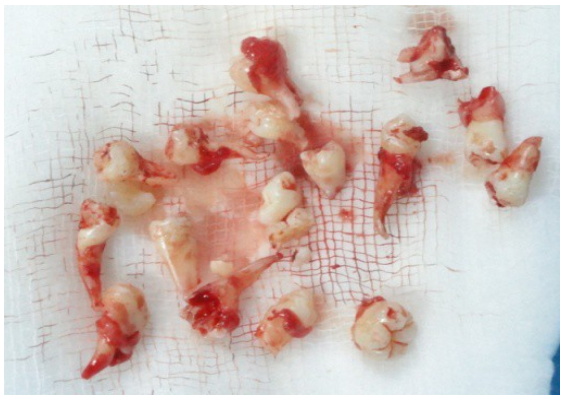
Radiological evaluation of skull, chest and jaw revealed frontal thickening, absence of bilateral clavicle ,absence of both gonial angle of mandible, multiple impacted supernumerary teeth mimicking premolars in shape, maxillary sinus seems as under developed. On screening orthopantomogram sixty four unerrupted supernumerary teeth were identified to be buried inside bone. The diagnosis is made by recognition of the both skeletal and dental components of this syndrome. No other family members had any features of this syndrome.

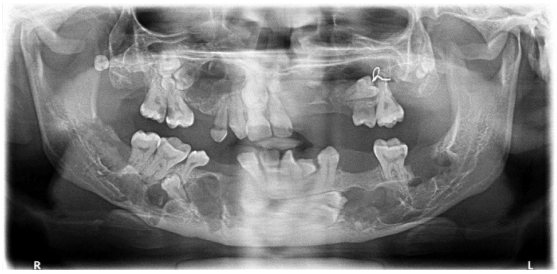




Management is mainly towards rehabilitation of occlusion by restoring the remaining retained teeth, surgical removal of impacted supernumerary teeth. Hypermobility of clavicle and problems with skull bones are not addressed as there is no expected complications.[5]

In this case surgical removal of supernumerary teeth were done under general anaesthesia, total removal of impacted teeth was not possible from maxilla and mandible. Post operative OPG showed some retained teeth (six). The patient further followed up for six months and referred for prosthodontic rehabilitation of occlusion and aesthetics.





Conclusion

Treatment of clediocranial dysplasia is mainly focused on reducing the dentofacial deformity and correcting the malocclusion. A coordinated treatment plan is required and generally involves removing some (but not necessarily all) supernumerary teeth. Recently there has been increasing use of multidisciplinary approach for treatment of these patients, utilizing the pedodontist, the orthodontist, and the oral surgeon.The retained deciduous teeth should be restored if they become carious,since their extraction does not necessarily induce eruption of permanent teeth. Those that seems to be forming dentigerous cysts or other pathologic entities or those that might interfere with orthodontic therapy and arch coordination are the ones indicated for removal. If a submucosal cleft exists, it can be corrected separately or at the time of orthognathic surgery, which may be indicated in some individuals. Follow-up orthodontic refinement and stabilization is necessary as is restorative and prosthetic dentistry for areas of carious or missing teeth. Uncovering of unerrupted teeth with planned attempts to guide eruption by orthodontic means cannot be expected to be successful because of the lack of cellular cementum. Life expectancy is normal.[7]

No treatment is directed towards skull and clavicular deformities, but the dental problems represent a major source of functional and esthetic morbidity. Without treatment, the retained deciduous dentition begins to rapidly deteriorate in late youth and early adulthood, leading to premature aged facial appearance.[1]

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