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CLEIDOCRANIAL DYSPLASIA: A CASE REPORT WITH LONG TERM FOLLOW-UP

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Abstract:

Aim: The purpose of this study was to evaluating a case of Cleidocranial dysplasia.

Background: Cleidocranial dysplasia (CCD) is a rare developmental disease with skeletal malformation affecting bones and teeth and is considered to be of autosomal dominant inheritance.

Case description

Here is a clinical case of a 7-year-old boy with a chief complaint of pain in posterior region. In initial evaluation we found that he has a few supernumerary teeth, during the period of follow up, increase in the number of supernumerary teeth was detected. It was 5 in 2009 and reached to 10 in 2015.Our probable diagnosis was cleidocranial dysplasia, afterword, we examined his mother and sibling. They displayed some signs of cleidocranial dysplasia. These findings confirm our presumed diagnosis.

The final treatment plan for this patient was pulpectomy and stainless steel crown for retained deciduous teeth and stainless steel crown for first permanent molars due to carious lesion, at last, in anterior region of maxilla, an esthetic nance was utilize for rehabilitation of phonetic and esthetic ability

Conclusion: These conditions of this disease have clinical significance to every dentist due to the involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth.

Key words: Cleidocranial dysplasia Delayed eruption Supernumerary teeth

Cleidocranial Dysplasia: a case report with long term follow-up.

Introduction

Cleidocranial dysplasia (CCD) is a rare developmental disease with skeletal malformation affecting bones and teeth and is considered to be of autosomal dominant inheritance. Individuals have been shown to be of shorter stature than unaffected relatives, also they have brachycephalic skull and bossing of the parietal and frontal bones .Vertical facial growth is decreased due to poor development of the alveolar bone (1). Other oral manifestations include prolonged retention of the primary dentition and delayed eruption of the permanent dentition, multiple supernumerary teeth and predilection for cyst formation involving the uneruptedteeth(2,3).

The absence of the clavicles, which occurs in 10 %of cases or the presence of hypoplastic clavicles allow the patient the movement of the shoulders up to the medial plan of the body without any discomfort, so the most typical CCD marker is extreme shoulder mobility, however is not always expressed. Nevertheless, the majority of craniofacial findings are age related and become obvious only during adolescence(4,5).

Cleidocranial dysplasia was first described by Pierre Marie and Paul Sainton in 1898, since then, over 1000 cases have been documented in the medical literature .it is caused by a mutation in runt-related transcription factor 2 gene (RUNX2), located on chromosome 6p21, have been identified as the cause of CCD. Being genetic in nature the disease may pass generation to generation as any other asset.As said before, In these patients maxilla is underdeveloped along with illformedparanasal sinuses. This condition is of clinical significance to every dentist due to the involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth(6,7,8,9,10).

Case Report

In 2009, a 7-year-old boy was referred to the pediatric department of dental clinic of Hamadan University of medical sciences with a chief complaint of pain in posterior region. A panoramic and two bitewing radiographs were prescribed for evaluation of developing dentition and carious lesions in posterior region. In radiologic assessment we noticed that he had a few supernumerary dental folliclesin his both jaws(4 in mandible in anterior and premolar region and 1 in left maxilla in premolar region)(figure 1). At that time, his teeth were treated and follow up sessions were advised.3years later, he attended again, He told that he hadn't had any teeth in anterior regions of his upper jaw since childhood after his milk teeth shed away(figure 2). His mother told that his smile was very unpleasant, and he was often teased at school, resulted in a psychological trauma to him while communicating with his classmates. He insisted onrehabilitation of his missing teeth and smile. Examination of the oral cavity revealed multiple over-retained deciduous teeth and some unerupted permanent teeth, particularly in anterior region of maxilla, only first

permanent molars were erupted. There was no evidence of eruption of maxillary and mandibular incisors. A panoramic and a lateral cephalometric radiograph were taken. On evaluating the panoramic radiograph the patient had more supernumerary teeth in his jaw, compared with the previous one (5 in mandible and 2 in maxilla) (figure 3). In cephalometric radiograph analysis it was revealed that this patient had hypoplastic maxilla and large mandibular body. The mandible has rotated clockwise that resulted in class I molar relationship and convex profile (figure 4). In consult with oral and maxillofacial surgery and orthodontics department, it was decided to operate an incision through fullthickness of gingival mucosa in order to accelerate the eruption of incisors and then an orthodontic was performed.

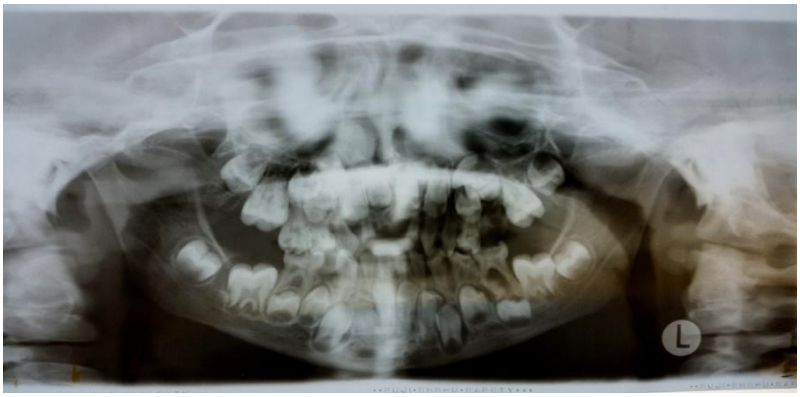

Fig 1: Panoramic radiograph shows a few supernumerary dental follicles in his both jaws.

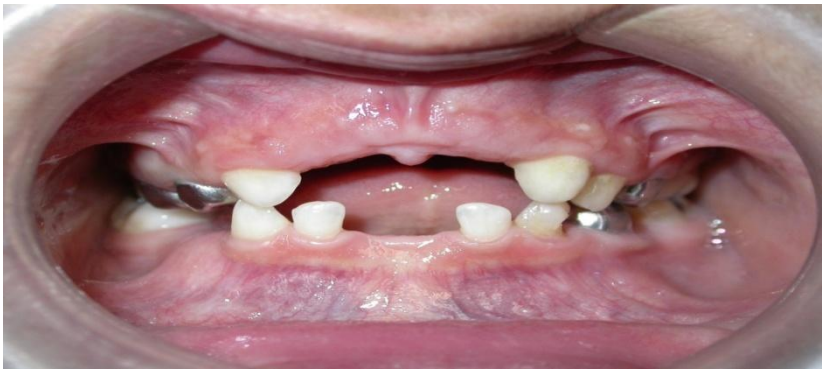

Fig 2: Photograph of Intra oral view of the patient.

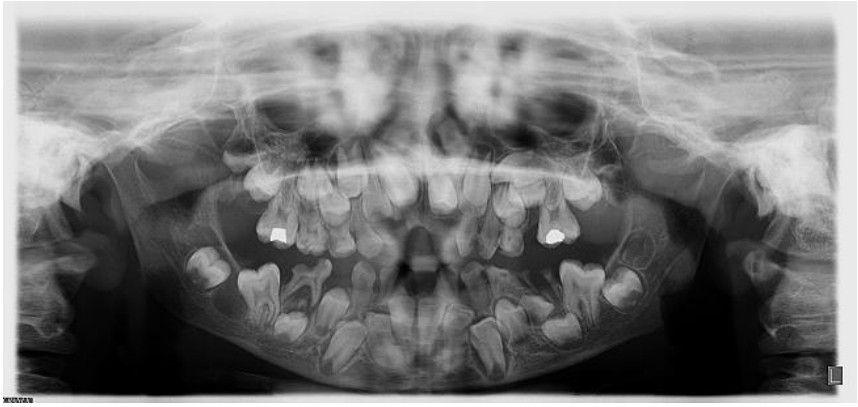

Fig 3: Panoramic radiograph shows that the number of supernumerary teeth has been increased.

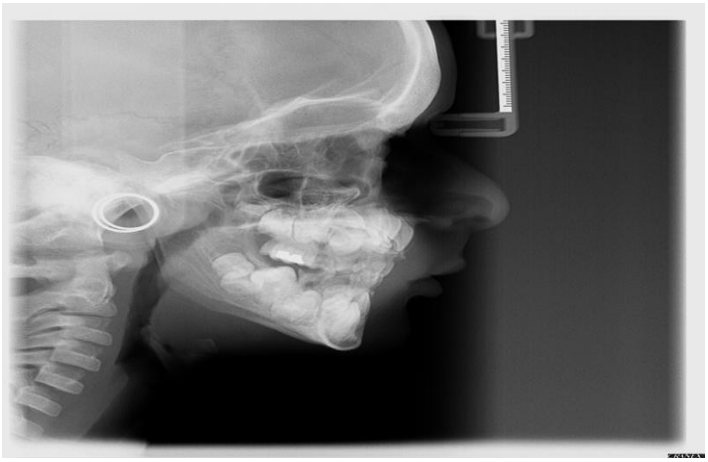

Fig 4: Lateral cephalogram reveals that this patient has hypoplastic maxilla and large mandibular body.

In follow up sessions, no movement of anterior teeth was observed, in clinical and radiographic assessment. In examination of whole body, The patient had short stature while his growth hormone rate was normal, he was then, asked to attempt to place his shoulders adjacent to each other to check for the incomplete or absence of clavicle bone formation and this attempt demonstrated more than normal mobility of the shoulder girdle(figure 5) . Upon re-examination of the face, it was found that he also had the symptoms of frontal and parietal bosselation(figure 5).

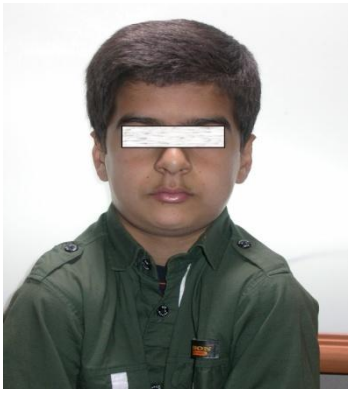

Fig 5: Frontal bossing, sloping of shoulders and approximation of shoulders towards each other.

We kept in mind that he might havecleidocranialdyplasia due to his clinical and radiographic symptoms. It was advised for complete surgical removal of supernumerary teeth and fixed orthodontic treatment. Because of psychological statement of the patient, he dodged the appropriate treatment and didn't attempt to attend for follow - up sessions.3 years later, the patient was attended again, and for eruption of permanent teeth, he was referred to another radiology center to take a cone Beam CT and new panoramicradiograph to assess the exact situation of his deciduous and permanent teeth meticulously. The number of his supernumerary teeth has been increased in comparison with 3 years ago (7 in mandible and 3 in maxilla)and roots of posterior deciduous teeth had not been resorbed(figure 6,7). However, at that time he was a 13-year old boy, he had only four permanent teeth in his oral cavity(first molars).

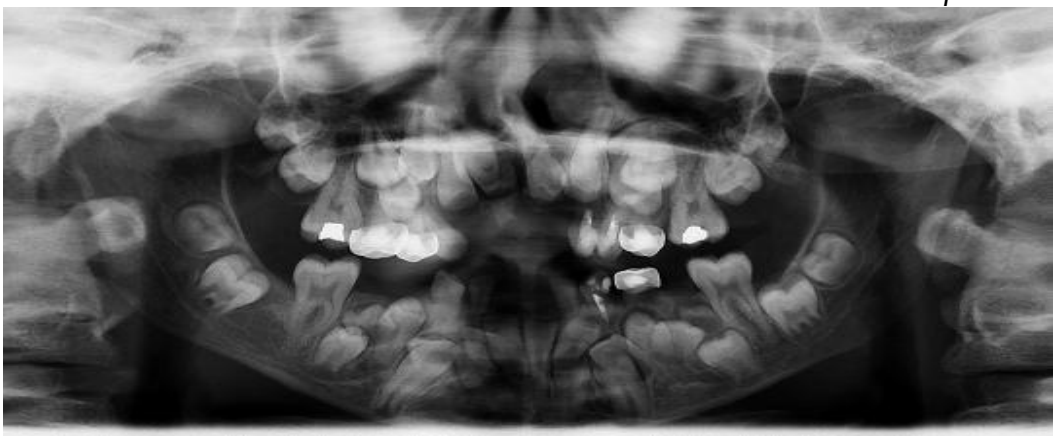

Fig 6: Panoramic radiograph after 3 years, shows that the number of supernumerary teeth has been increased again.

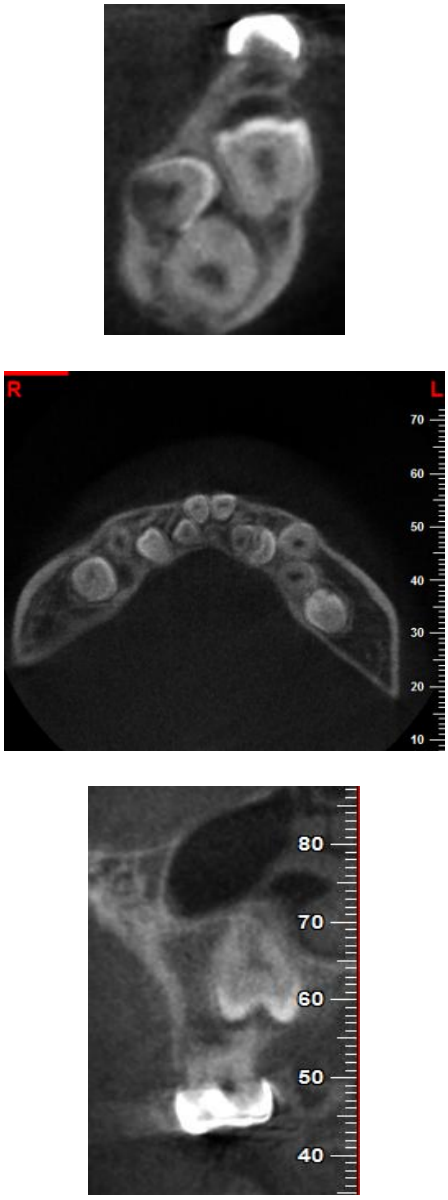

Fig 7: Cone beam CT images (A, B and C)Reconstructed panoramic (a) and cross sectional images(b and c) clearly demonstrate the position of impacted teeth and its relation to adjacent teeth, note that roots of the posterior deciduous molar have not been resorbed.

Our probable diagnosis was cleidocranial dysplasia, afterword, we examined his mother and sibling. They displayed some signs of cleidocranial dysplasia such as abnormal shoulder and skull, a lot of supernumerary teeth and so on (figure 8). These findings confirm our presumed diagnosis.

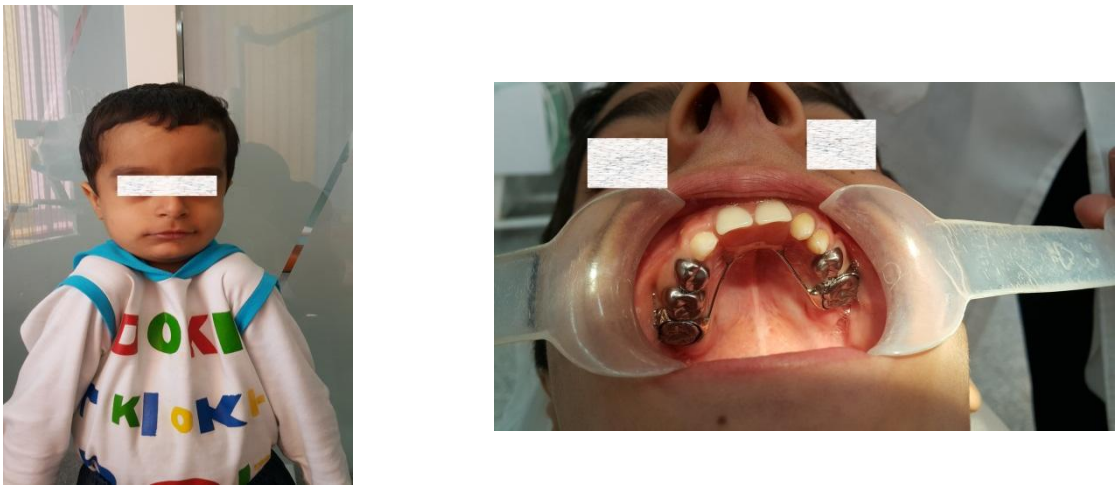

Fig 8: Younger brother: Extraoralphotograph(note the approximation of shoulders).

The final treatment plan for this patient was pulpectomy and stainless steel crown for retained deciduous teeth and stainless steel crown for first permanent molars due to carious lesion, at last, in anterior region of maxilla, an esthetic nance was utilize for rehabilitation of phonetic and esthetic ability(figure 9).

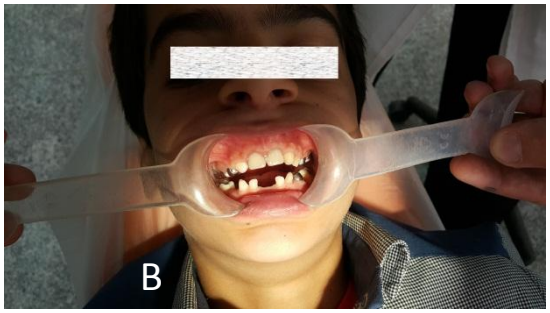

Fig 9: (A and B) Estheticnance was utilize for rehabilitation of phonetic and esthetic ability.

Discussion

The molecular defect in CCD is situated at the chromosome 6p21. Although CCD is relatively infrequent, it has a wide geographic distribution presumably due to in progress random occurrence of new mutations in the determinant gene. In this sequence of events, there is no biological pressure against autosomal dominant transmission from generation to generation and a chance of mutation or a tremendous effect can be sustained in a particular population(1)

Patients with CCD have short stature and have frontal, parietal and occipital bossing leading to bulging calvarias. There is a partial or complete absence (in about 10% of cases) of the clavicle result in hypermobility of shoulders,

open fontanelles, wormian bones, a wide pubic symphysis, short middle phalanges of the fifth fingers, and various vertebral and dental abnormalities. Dental manifestations are underdeveloped maxilla, relative mandibular prognathism, retained primary dentition, multiple impacted permanent dentition, delayed eruption of permanent teeth, multiple supernumerary teeth, crown and root abnormalities, cyst formation around impacted teeth, and a high palate that are seen to be consistent features of cleidocranial dysplasia. Various scientific theories has been postulated with respect to the etiology of non-eruption of permanent teeth, such as lack of cellular cementum, defectiveness in post cementum formation, presence of thick connective tissue between oral epithelium and dental follicle, delayed tooth formation and maturation(2,4) our case demonstrated some of cited symptoms such as short height, hypermobile shoulder, retained deciduous teeth, unerupted and impacted permanent teeth, unresorbed root of deciduous teeth, hypoplastic maxilla, large mandibular body and familial history which confirm our diagnosis.

Treatment planning for this group of patients is difficult, Zi-Jian Li etal(1), adopted surgical exposure of the unerupted permanent teeth with orthodontically guided eruption as their method of choice. This allowed the patient to retain his own teeth and avoided the need for a prosthesis that would have to be maintained or replaced several times during his lifetime. The patient was satisfied with the esthetic and functional aspects of the treatment. This finding indicates that some of these patients have normal cementum and other factors are responsible for lack of eruption. These factors comprise the anatomical obstacles like supernumerary teeth, thick overlying gingiva and so forth. On the other hand, Garg R et al(8) and Butterworth (9)believe that despite good surgical exposure and active orthodontic traction at an appropriate age, permanent incisors in CCD do not always erupt, hence they applied prosthetic appliances to rehabilitate the missing teeth.In the present study, surgical exposure of maxillary incisors was not effective despite active orthodontic forces, and the incisors remained fixed in the alveolus, so support this theory that insome of these patients cementum is abnormal and lack of cellular cementum is present(9).

The other factor that guided us to confirm the diagnosis of this disease was presence of familial relevance in this case, so that two members of his family (mother and one brother) suffered from some of these present medical conditions. It is accordance with the findings of Marthur A et al(10) that studied manifestations three siblings which had cleidocranial dysplasia. Although Bharti K et al (2) and Alves N et al(5) did not find any familial relevance and not report the existence of direct ancestors or descendents who presented any clinical characteristic of cleidocranial dysplasia. The characteristic feature of this case was the overwhelming increase in the number of supernumerary

teeth during the period of follow up. It was 5 in 2009 and reached to 10 in 2015. It is noteworthy and the future follow up is recommended.

At last, the suggested treatment for dental and maxillofacial complications of cleidocranial dysplasia is:

1 .Fabrication of dentures over the unerupted teeth, or

2. Removal of teeth as they erupt, for very little bone structure would be left if supernumerary, impacted and unerupted teeth were all extracted at once.

3. Orthodontic treatment for their maxillofacial problem and alignment of their teeth(8)

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