Cleidocranial dysplasia associated with the dentigerous cyst: review of the literature and report of the clinical case

Displasia cleidocraniana associada a cisto dentígero: revisão da literatura e relato de caso clínico

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Abstract

Cleidocranial dysplasia is a developmental anomaly of the skeleton and teeth. It can be inherited as an autosomal dominant characteristic with high penetrance and variable expressivity. It occurs with equal frequency in both genders and races. The clinical appearance of cleidocranial dysplasia is pathognomonic. The stature is mildly to moderately shortened, with neck appearing long and narrow and the shoulders markedly drooped. Maxillary hypoplasia gives the mandible a relatively prognathic appearance, although some patients may show variable mandibular prognathism due to increased length of the mandible in conjunction with short cranial base. The head and neck features are very variable. Herein, we are described one cleidocranial dysplasia case, in an 84 year female patient, with the dentigerous cyst, which she were not kwon the diagnosis of the her congenital syndrome.

Descriptors: Cleidocranial dysplasia; Dentigerous cyst; Mandibular injuries; Tooth, supernumerary

Resumo

A displasia cleidocraniana é uma alteração de desenvolvimento do esqueleto e dos dentes. É uma desordem autossômica dominante com alta penetrância e expressibilidade variada. Ocorre em igual frequência em ambos os gêneros e raças. Suas características clínicas são patognomônicas. Apresenta estatura baixa a moderada, com aparência do pescoço longo e os ombros caídos. Hipoplasia da maxila gera um aparente prognatismo. As características clinicas em cabeça e pescoço são muito variadas. Este relato descreve um caso de uma paciente de 84 anos com displasia cleidocraniana associada a cisto dentígero a qual não sabia ser portadora de tal síndrome congênita.

Descritores: Displasia cleidocraniana; Cisto dentígero; Traumatismos mandibulares; Dente supranumerário

Introduction

Cleidocranial dysplasia is a developmental anomaly of the skeleton and teeth. It was first described in 1897 by Marie and Sainton, who termed the condition cleidocranial dysostosis[1]. It can be inherited as an autosomal dominant characteristic with high penetrance and variable expressivity. Few cases of recessive forms have been reported. About one third of the cases is sporadic and appears to represent new mutations[2-4]. It occurs with equal frequency in both genders and races with a prevalence of less than 1 per million[5].

The clinical appearance of cleidocranial dysplasia is pathognomonic. The stature is mildly to moderately shortened, with neck appearing long and narrow and the shoulders markedly drooped. Primarily affects the skull, clavicles, and dentition. The facial bones and paranasal sinuses are hypoplastic, giving the face a small and short appearence. This is result of hypoplasia of the maxilla, a brachycephalic skull, and the presence of frontal and parietal bossing. The bridge of the nose may be broad and depressed, and hypertelorism is present[5-10].

Complete or partial absence of clavicular calcification, with associated muscle defects, results in hypermobility of the shoulders, allowing for variable levels of approximation in an anterior plane. Other bones also may be affected, including the long bones, vertebral column, pelvis, and bones of the hands and feet. Hemivertebrae and posterior wedging of the thoracic vertebrae may contribute to the development of kyphoscoliosis and pulmonary complications[7-8,10].

Maxillary hypoplasia gives the mandible a relatively prognathic appearance, although some patients may show variable mandibular prognathism due to increased length of the mandible in conjunction with short cranial base. The palate is narrow and highly arched, and there is an increased incidence of submucosal clefts and complete or partial clefts of the palate[11-12].

The head and neck features are very variable[12-13]. Herein, we are review the clinical features of the syndrome in maxillaries bones, was well described one cleidocranial dysplasia case, in an 84 year female patient, with the dentigerous cyst, which she were not kwon the diagnosis of the her congenital syndrome.

Case report

A 84-year-old female was referred to the Oral Clinic, Dental School, University Paulista, Goiânia-GO, Brazil, for dental treatment. The patient complained of dysphagia and noted that she had experienced difficulty swallowing and difficulty to use the dental prosthesis for approximately two weeks. Her daughter reported that patient complained about the eruption of teeth in maxillary bone and pain in mandible parasymphysis region. During anamnesis it was observed that patient had a significant hearing loss and it was always required the presence of his daughter during the dialogues and communication. Previous family medical history was noncontributory.

On physical examination it was observed short height and stature, maxillary hypoplasia, delayed in teeth eruption, shrugged shoulders and narrow chest, hypermobility of the shoulders both in the patient and her daughter (Figures 1 and 2). She also had a prominent frontal boss and prognathic appearance. With a clinical diagnosis of a cleidocranial dysplasia, the head and thorax radiographic exams were performed. Radiographicaly, it was possible observed the bilateral hypoplasia of clavicles and impacted supernumeraries teeth, which confirmed the clinical diagnosis the cleidocranial dysplasia (Figures 3 and 4). Also, it was observed a cystic formation in mandible parasymphysis and noted the possibility of mandible fracture. Oral clinical examination revealed an expansive mass in the mandibular region around the cyst.

The surgical procedure to remotion of supernumerary teeth and an excisional biopsy of the cystic lesion was taken while the patient was under general anesthesia. Microscopic examination of the hematoxylin-eosin (H&E) specimen showed the presence of thin fibrous wall

lined by keratinized stratified squamous epithelium associated with chronic inflammation, confirming the diagnosis of dentigerous cyst.

The patient and his daughter were referred to the Clinic Hospital for further management and follow-up of their systemic disease. The patient was followed up and 12 months after surgery there is no sign of recurrence and she not complained pain, and it was not possible to observe the presence of supernumerary teeth exposed in oral cavity (Figure 5).

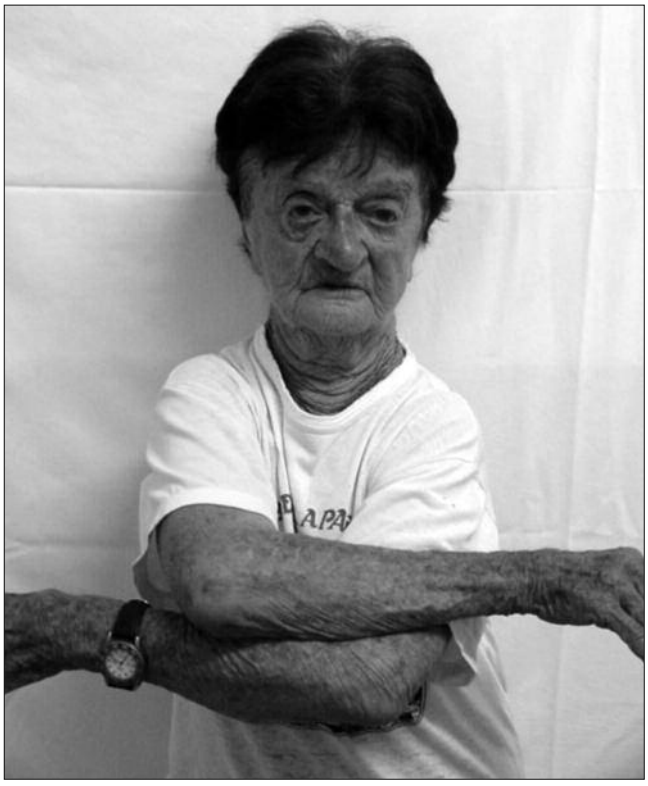

Figure 1. A 84 year-old female patient. It was possible to observe the hypermobility of the shoulders, allowing for variable levels of approximation in an anterior plane

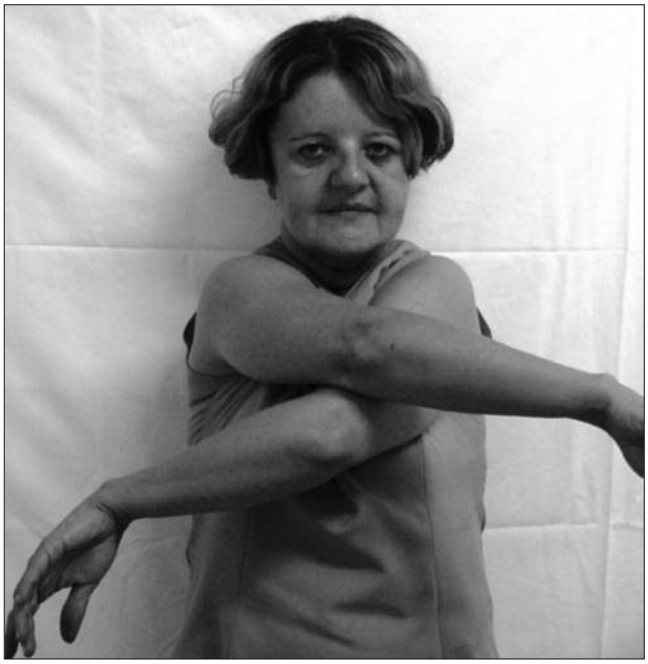

Figure 2. The daughter of the patient. She also present the mildly shortened, with neck appearing long and narrow and the shoulders markedly drooped

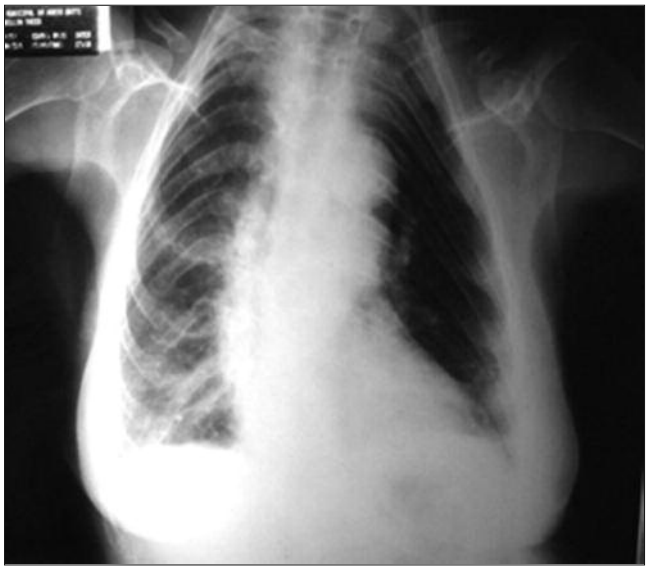

Figure 3. Radiographic exam of the thorax. It was possible to observe the bilateral hypoplasia of clavicles

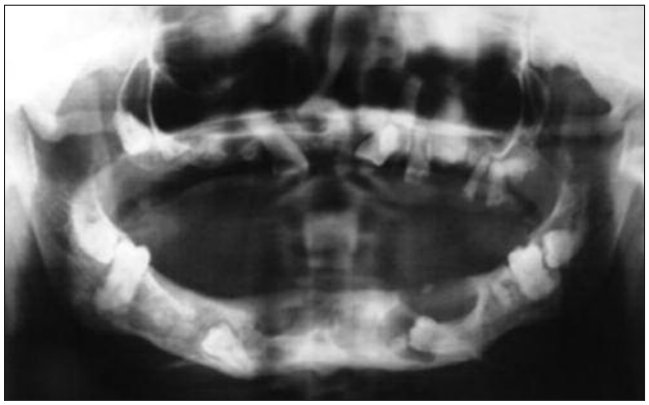

Figure 4. Radiographic oral exam. It was possible observed the impacted supernumeraries teeth and a cystic formation in mandible parasymphysis

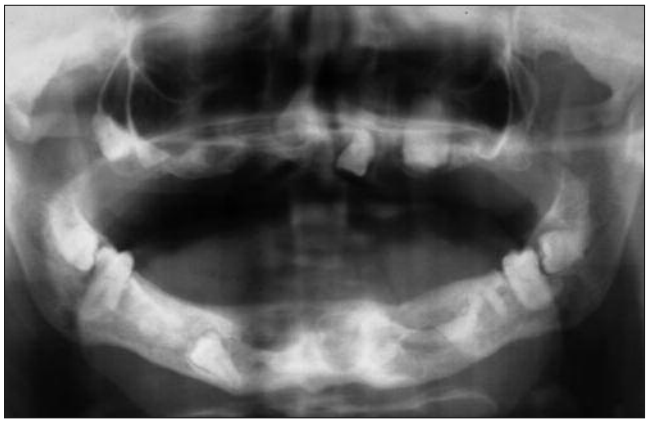

Figure 5. The patient was followed up and 12 months after surgery there is no sign of recurrence

Literature review and Discussion

Patients with cleidocranial dysplasia show prolonged retention of the primary dentition and delayed eruption of the permanent dentition. Extraction of primary teeth does not stimulate eruption of underlying permanent teeth. A study of teeth from patients with cleidocranial dysplasia revealed a paucity or complete absence of

cellular cementum on both erupted and unerupted teeth. It is postulated that failure of cementum formation may be due to mechanical resistance to eruption by the dense alveolar bone overlying the unerupted teeth[11-14].

Often unerupted supernumerary teeth are present and considerable crowding and disorganization of the developing permanent dentition may occur. Unerupted supernumerary teeth are frequently present in all regions. Only one supernumerary per normal tooth is generally noted. The unerupted teeth develop most commonly in the anterior maxilla and bicuspid regions of the jaws. Many resemble bicuspids, and these unerupted teeth, may developed dentigerous cysts[10-11,13,16-17].

Formation of supernumerary teeth is due to incomplete or severely delayed resorption of the dental lamina, which is reactivated at the time of crown completion of the normal permanent dentition. The over retention of deciduous teeth, failure of eruption of permanent teeth, numerous supernumerary teeth, and maxillary hypoplasia result in severe malocclusion[15-16,18-20].

There is no specific treatment for patients with cleidocranial dysplasia. Genetic counseling is most important. Protective headgear may be recommended while fontanels remain patent. The current mode of dental therapy combines surgical intervention with orthodontic therapy. Early surgical exposure of unerupted teeth has resulted in stimulation of cementum formation and eruption of the dentition with normal root formation[14-15, 17-20].

Conclusion

The dentist should be aware that oral involvement in cleidocranial dysplasia is common and frequently the first sign or complaint of the disease. Cleidocranial dysplasia is most relevant to dentistry because lesions may involve especially the head and neck, typically the teeth and maxillary bones. The patient reported herein received diagnosis of cleidocranial dysplasia due to oral complaint. It was confirmed the value of dentistry on systemic diseases diagnosis.

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