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Cleidocranial Dysplasia – A Late Diagnosis

Abstract: Cleidocranial dysplasia is a rare congenital disorder of bone characterized by abnormalities of clavicle, skull and dentition. There are features which are pathognomic for this condition and diagnosis can usually be made early in life. This report presents a case that was diagnosed very late in life, with a resultant compromised dental state, and discusses current treatment modalities and the differential diagnosis with another condition with which this was mistaken.

Clinical Relevance: Delayed eruption of permanent teeth must be diagnosed and investigated early so that it can be managed appropriately.

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Cleidocranial dysplasia (CCD) is a rare congenital disorder of bone characterized by abnormalities of the clavicles, skull and jaw, as well as by the occasional limitation of growth of long bones. The oral manifestations are failure of eruption of the secondary dentition and multiple unerupted supernumerary teeth.[1]

Early publications suggested that cleidocranial dysplasia (also known as cleidocranial dysostosis) was recognized as early as 1760,[2,3] although through descriptions in literature such individuals suffering from the disease or one very similar may have been described earlier.[4,5,6] For example, in the Homeric epics the Iliad and Odyssey, the character Thersites was described as:

'bandy legged and lame in one foot; And his two shoulders were rounded, stooping together over his chest; And above them his head was warpen, and a scant stubble grew thereon.

The description of Thersites' head and legs are not diagnostic for CCD, but Carter[5] felt that the ability to bring the shoulders together in front of the chest is found only in this disease.

In 1897, Marie and Sainton[7] appear to be the first to use the term 'cleidocranial dysostosis' to describe individuals with the following features:

- Aplasia or hypoplasia of one or both clavicles;
- Exaggerated development of the transverse diameter of the cranium;
- Delayed ossification of fontanelles.

The associated problems of failure in spontaneous eruption of permanent teeth in patients with CCD can be attributed to the persistence of the primary dentition and also as a result of diminished bone resorption.[8,9]

Historically, a number of methods of managing the dental problems associated with CCD have been described. These include:[10]

- Combined surgical/prosthetic treatment – involving the removal of all persisting primary teeth and relying on the permanent teeth to erupt spontaneously. This often results in eruption of only a few permanent teeth. Subsequently, all the erupted and retained teeth can be removed and full dentures fabricated.
- Combined surgical/orthodontic treatment – involving the exposure of the permanent teeth after which they are orthodontically extruded. This primarily involves patient groups of 15 to 36 years of age and a mean treatment time of 8 years. Varying degrees of success were recorded.
- Autotransplantation where unerupted teeth are surgically removed and replanted in associated areas.

More recent treatment methods described include:[10]

- Early diagnosis with possible removal of the primary and supernumerary teeth before the completion of root formation of the permanent teeth. This, along with possible surgical exposure, improves the erupting potential of the teeth.
- Multidisciplinary approach involving paedodontist, oral and maxillofacial surgeon, orthodontist and restorative dentist.

The objective of such treatments is to retain as much of the permanent dentition as possible, with teeth positioned in their most useful sites if restorative intervention is

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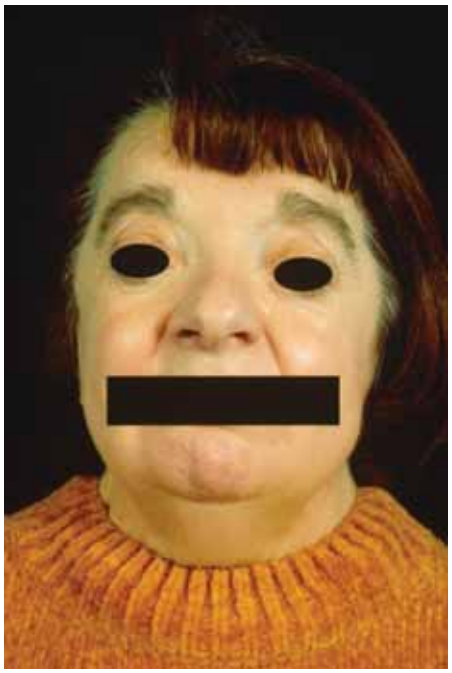

Figure 1. Typical facial features.

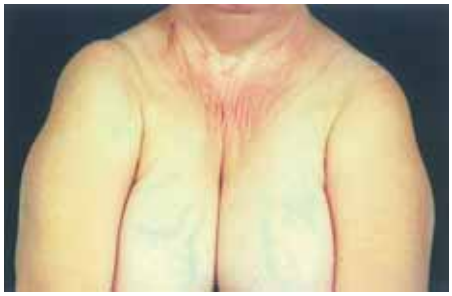

Figure 2. Ability to approximate shoulders.

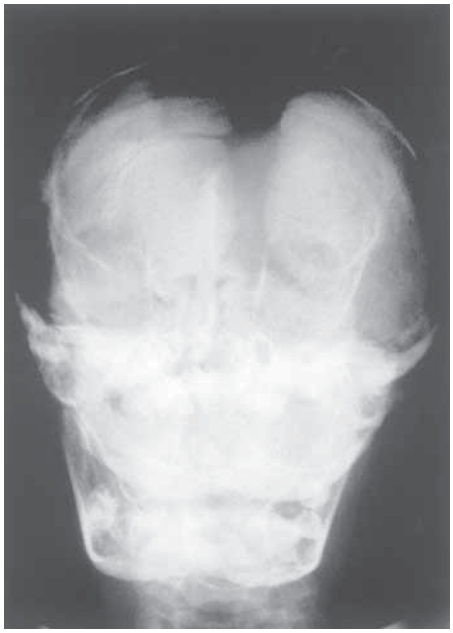

Figure 3. PA skull radiograph.

necessary.

Such strategies underline the importance of early diagnosis and referral of CCD patients for multidisciplinary management of emerging dental problems

This paper describes a patient who was not diagnosed early, resulting in a compromised dental outcome in the long term.

Clinical presentation

A 57-year-old female was referred by her General Dental Practitioner (GDP) to the Oral and Maxillofacial Surgery clinic at the hospital dental services. She had presented to the GDP complaining of a swelling on the left side of the mandible, which was making denture wear uncomfortable. The GDP obtained an OPT which revealed a full, unerupted dentition.

The patient revealed on questioning that she had seen her dentist as a teenager and asked him to remove her remaining erupted teeth, as they were spaced and were a source of embarrassment to her. Full clearance of the erupted teeth was carried out and complete upper and lower dentures fitted.

There was no relevant family history other than the patient's claim that she was told that she had a 'calcium' problem. No more details regarding this claim could be ascertained from the patient.

In the past, the patient reported experiencing fractures of the right elbow (1988), right wrist (1990), and left upper arm (1995). In 1995, subsequent to the fracture of her left upper arm, she was referred to a Skeletal Dysplasia Clinic (SDC) where a diagnosis of Melnick-Needles Syndrome (MNS) was made. The patient could not recall whether any dental history was obtained by the clinic and, as she required no further treatment, she was discharged.

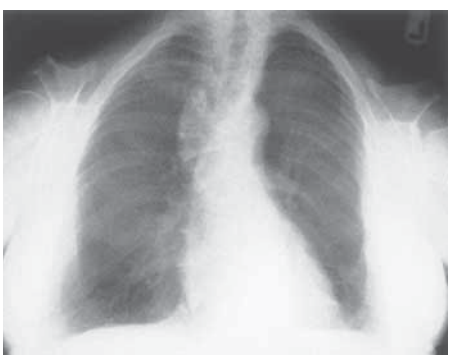

Figure 4. Chest radiograph.

General examination revealed a short stature and frontal and parietal bossing. Other facial features included (Figure 1):

- Hypertelorism;
- Depressed and widened nasal bridge;
- Diminished midface height;
- Brachycephalic head.

The patient was able to approximate her shoulders in an anterior plane (Figure 2). Intra-oral examination revealed a few erupting teeth and a fluctuant swelling of the gingiva in the left lower molar region. The patient indicated that the swelling was making denture wear difficult. Aspirate of the swelling showed cystic fluid with apparent cholesterol crystals.

The following radiographs were taken which revealed:

PA Skull (Figure 3):

- Persistence of fontanelles;
- Presence of metopic suture;
- Underdeveloped paranasal sinus;
- Presence of multiple wormian bones.

Chest X-ray (Figure 4):

- Complete absence of clavicles;
- Narrow cone-shaped thorax;
- Double curve pattern scoliosis.

OPT (Figure 5):

- Multiple permanent and supernumerary teeth;
- Narrow ascending ramus;
- Slender coronoid process and curved distally;
- 3 x 3.5 cm circumscribed radiolucent area with sclerotic border in relation to $\overline{|6\ 7}$.

Diagnosis

Based on the clinical and radiographic evidence, a diagnosis of cleidocranial dysplasia was made.

The cyst was enucleated along with the $\overline{|6\ 7}$ under general anaesthesia and the specimen sent for histopathology and tooth histology. The postoperative period was uneventful and the patient was discharged on the same day with necessary instructions. The patient was reviewed 2 weeks later when she reported no significant problems.

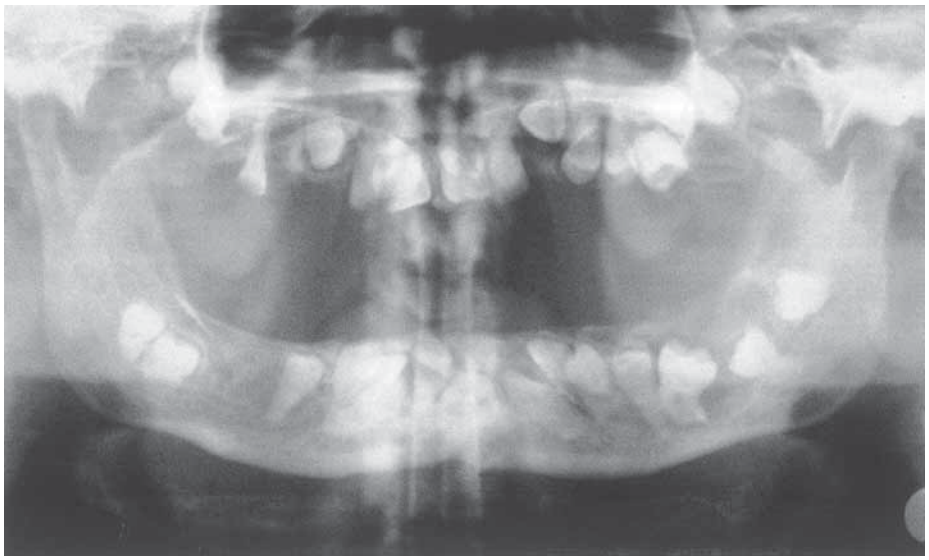

Figure 5. Orthopantomogram showing unerupted teeth.

Microscopic examination of the cystic material revealed a dentigerous cyst. Histopathology of the extracted teeth revealed absent cementum in places but thin elsewhere. The cementum present was acellular.

Discussion

Generalized delayed eruption of permanent teeth is attributable to a number of factors including:[11]

- Cleidocranial dysplasia;
- Cretinism;
- Rickets;
- Hereditary gingival fibromatosis.

Current guidelines suggest that prolonged retention of deciduous teeth should be investigated.[10] It would appear that the cause of retention of this patient's deciduous dentition, and failure of the permanent successor to erupt, was not investigated at the time of the dental clearance when she was 15 years old.

It was not until she attended a Skeletal Dysplasia Clinic (SDC), when aged 51 years, that the possibility of an underlying pathological condition was considered. This was the result of a history of multiple long-bone fractures, some 36 years after the presence of retained deciduous teeth were apparent. At the SDC, cleidocranial dysostosis was considered in the initial differential diagnosis, together with Melnick-Needle Syndrome(MNS).

Melnick-Needle Syndrome is an X-linked dominant bone dysplasia described in 1966.[12] The clinical and radiographic features are:

- Hypertelorism;
- Short stature;
- Outstanding nose and receding chin;
- Delayed closure of anterior fontanelle;
- Generalized bone dysplasia;
- S-shaped tibia;
- Ribbon appearance of ribs;
- Pelvic bone abnormalities – coxa valga.

Cleidocranial dysplasia is a rare congenital disorder of bone characterized by abnormalities of clavicle, skull and dentition and other bones. This condition may be inherited as an autosomal dominant character in either gender. It has also been known to appear spontaneously as a result of mutation. It has no racial predilection and individuals with this condition usually have normal intelligence.

The aetiology seems to be a mutation in the CBFA1 gene on chromosome 6p21 and this mutation is thought to be a submicroscopic deletion. The CBFA1 gene controls osteoblast differentiation from precursor cells and is essential for bone formation – both membranous as well as endochondral.

Although there are a number of common features between CCD and MNS, the complete or partial absence of the clavicle is usually indicative of cleidocranial dysplasia. In addition, MNS is not associated with any dental problems. Unfortunately, it would appear that no dental history or radiographic evaluation was obtained when the patient attended the SDC at a time when, clinically, she was edentulous. This is unfortunate, since the presence of multiple unerupted teeth would have probably led to a diagnosis of CCD and the instigation of an appropriate management regime. In particular, clinical and radiographic monitoring of the unerupted teeth could have occurred and, possibly, in respect to cystic changes, active intervention initiated earlier. In addition, appropriate intervention may have been carried out in respect to managing the unerupted teeth to benefit any possible restorative intervention.

Dentigerous cysts are commonly associated with teeth with delayed eruption and have been described previously in CCD.[8,13] Histopathology of the extracted teeth revealed complete lack of cellular cementum. Rushton first noted this in 1956 and suggested that this may be an additional dental defect commonly associated with CCD.[14] This finding has been confirmed by other authors.[15,16]

This case report illustrates the importance of investigating any instances of unusual retention of primary dentition and obtaining a complete dental history when considering all patient groups, particularly those with potential skeletal pathologies. Investigations carried out at the appropriate juncture could have ensured diagnosis of this condition early with the patient benefiting from the most appropriate long-term management and monitoring.

Summary

Cleidocranial dysplasia is a rare congenital disorder of bone characterized by abnormalities of clavicle, skull and dentition and other bones. Partial or complete absence of clavicles, along with multiple unerupted teeth, strongly suggests this condition. Delayed eruption of dentition must therefore be thoroughly investigated so that appropriate treatment and management can be instituted.

Acknowledgement

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CPD ANSWERS

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1. B, D
2. A, B, D
3. A, B, C, D
4. A, B, C
5. A, B, D
6. A, C
7. A, B, D
8. B, C, D
9. B, C, D
10. A, C, D