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Dental clearance unsuccesful: cleidocranial dysplasia diagnosed at a relief of pain clinic

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Abstract

A 55-year-old woman presented to Whangarei Base Hospital Emergency Department with a mandibular swelling adjacent to her unerupted tooth 48. She had never had dental radiographs taken, having had a dental clearance 20 years earlier. She wore full dentures. A panoramic dental radiograph revealed 44 unerupted teeth. Based on the clinical, oral and radiographic examinations a diagnosis of cleidocranial dysplasia (CCD) was confirmed. The patient had not been diagnosed with CCD prior to her hospital visit.

Treatment included staged extractions of superficial mandibular supernumeraries and enucleation of the mandibular cyst, attempting to avoid pathological fracture. This would be followed by a six-month review and then annually as required. The case outlines the value of dental radiography in dental practice. Little is known about the prevalence of CCD in New Zealand and this is an area where future research could be conducted.

Introduction

Cleidocranial dysplasia (CCD), also known as cleidocranial dysostosis, or Marie and Sainton disease, is a rare congenital defect of autosomal dominant inheritance (Sekerci *et al*, 2013). The condition is characterised by clavicular aplasia or deficient formation of the clavicles, delayed and imperfect ossification of the cranium, moderately short stature, and a variety of other skeletal abnormalities (Reddy and Parmar, 2010). The ability to approximate the shoulders anteriorly is related to clavicular aplasia and is the classic diagnostic sign of the disorder. The gene responsible for cleidocranial dysplasia has been mapped to the short arm of chromosome 6, core binding factor alpha-1 (CBFA1). The syndrome can be caused by a mutation in the transcription factor CBFA1 which controls differentiation of precursor cells into osteoblasts and is therefore responsible for membranous as well as endochondral bone formation, hence the delayed ossification of the skull, pelvis and clavicles (Mundlos *et al.*, 1997).

Prevalence statistics are lacking, with some authors suggesting one per million with complete penetrance and variable expressivity (Cooper *et al.*, 2001). However, this condition is likely to be under diagnosed due to the lack of medical complications when compared to other skeletal abnormalities. It may be discovered at any age, but the cranial deficiencies may be noticed at birth. Both sexes are affected equally and the problem often appears in successive generations.

Abnormalities of the dentition present the most significant symptoms of CCD. Classical dental manifestations include retention of the primary teeth, impaction or delayed eruption of the permanent teeth and an irregular number of supernumerary teeth. Authors on CCD have suggested that supernumerary teeth form as a result of re-activation of remnants of the dental lamina left unresorbed during odontogenesis (Jensen and Kreiborg, 1990). The following case report is an incidental finding of a patient with CCD in the Northland region of New Zealand (NZ). The report highlights some of the difficulties encountered during patient management, treatment planning and surgical treatment. It also features an interesting cultural belief associated with CCD.

Case Report

A 55-year-old woman presented to the emergency department at Whangarei Base hospital with a mild right sided lower facial swelling. The swelling had been present for five days and it was due to a dental infection. The patient was a current smoker with high blood pressure, but no other significant medical history. She was given analgesia and antibiotics and referred to the oral health department for assessment and treatment as required. On presentation to her dental appointment she described having had a full dental clearance approximately 20 years before. She also reported that she had complete upper and lower full dentures, but was unable to wear her lower denture due to the swelling. Intraoral examination noted root-like fragments in the anterior maxilla and mandibular region. The swelling was in the buccal sulcus adjacent to the tooth 48 region. Suspecting retained roots may have been the cause of her swelling, she was sent for a panoramic dental radiograph (PDR) to help determine the origin of the swelling and whether there were any further retained roots. The patient reported to have never had any dental X-rays taken before.

Figure 1 shows the PDR which revealed approximately 44 unerupted and impacted supernumerary teeth. The radiologist's report stated "Full dental clearance unsuccessful". An area of concern was the large multilocular cyst-like appearance in the body of the left mandible. Having showed the patient the film she mentioned it was due to a family myth based on a Maori legend and that her relatives have similar 'bird like features' and extra sets of teeth. The patient had a relatively short and stubby stature with a brachyfacial profile. She explained that she did not have any collar bones (clavicles) and her skull cap (cranial sutures)

had never formed or hardened. She also revealed her malformed fingers and toe nails, which were photographed with her consent (Figures 2, 3, and 4).

Based on the clinical, oral and radiographic examination a diagnosis of CCD was confirmed. The patient was not aware of the syndrome, which was discussed with the patient and she was referred to the Oral and Maxillofacial Surgery outpatient clinic for treatment as required. The patient was given additional pain relief and antibiotics to continue to reduce the swelling and discomfort.

The patient was assessed in the Maxillofacial Clinic and a cone beam CT image was ordered (Figure 5). The treatment plan had two main objectives:

- Reduce the risk of further infection (swelling, pain etc)
- Reduce the risk of a pathological fracture of the left mandible (slow growing cyst)

Treatment would include staged extractions of the superficial mandibular supernumeraries and enucleation of the mandibular cyst while attempting to avoid pathological fracture. This would be followed by appropiate review. The maxillary supernumeraries did not present with any significant pathology, so no treatment was indicated.

The first stage of extractions under general anaesthesia proved difficult due to the orientation of the supernumeraries. Bilateral, conventional buccal and lingual flaps were raised in the mandible, bone was removed and a total of seven supernumerary teeth were extracted. The cyst within the left mandible was enucleated and post-operative instructions given. A review one week later was unremarkable and normal soft tissue healing had occurred. Histology results reported an odontogenic cyst.

The patient presented for a six-month review with no complaints of discomfort, pain or recurrent infection. Her concern was the ill-fitting mandibular complete denture, for which a reline or a new prosthesis was advised. A second PDR showed evidence of bony

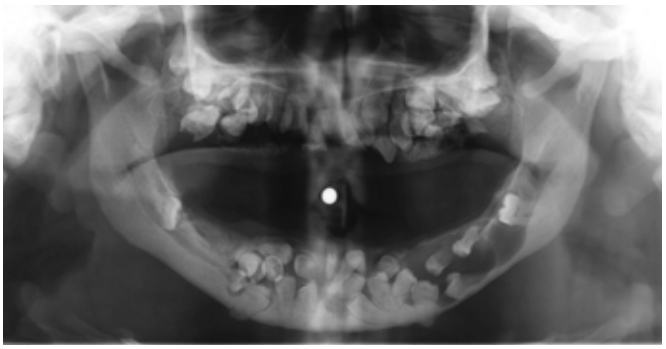

Figure 1. Panoramic dental radiograph

healing where the superficial supernumeraries had been removed and also in the mandible where the cyst was enucleated. Annual reviews will be required to further monitor bony healing or any pathological changes.

The patient reports that her mother and sister have similar dental traits and she has two siblings, a brother and a sister. The males in her immediate family did not have any known dental anomalies. Attempts to follow up the patient's other relatives were challenging due to cultural sensitivity (Maori legend) and pre-determined beliefs (family myth) about the CCD syndrome.

Discussion

Dental findings in patients with CCD are characterized by abnormal dentition, including malocclusion, irregular forms of dentition, wide spacing in the lower incisor area, supernumerary tooth germs, parallel-sided ascending rami, cysts of the gingiva that usually form around extra teeth and increased odontogenesis leading to excessive number of supernumerary teeth (Sekerci *et al.*, 2013). The suggested aetiology for the alarming number of supernumerary teeth is the incomplete or severely delayed resorption of the dental lamina, which is then reactivated at the time of crown completion in the normal permanent teeth (Reddy and Parmar, 2010).

Several approaches have been reported for the dental management of CCD. No treatment at all was very common in the past (Becker *et al.,* 1997). The option of a full clearance and dentures has also been suggested (Winter, 1943). However, some authors regard this approach as too invasive, due to the significant bone loss experienced after removal of teeth in a patient already deficient in alveolar bone (Daskalogiannakis *et al,* 2006). Pusey and Durie (1943) suggested removal of only the erupted teeth and the use of a removable prosthesis to minimize alveolar bone loss. However, it has been shown that subsequent eruption of retained teeth can require further surgery and modification of the prosthesis (Bekker *et al.,* 1997). The current treatment described by

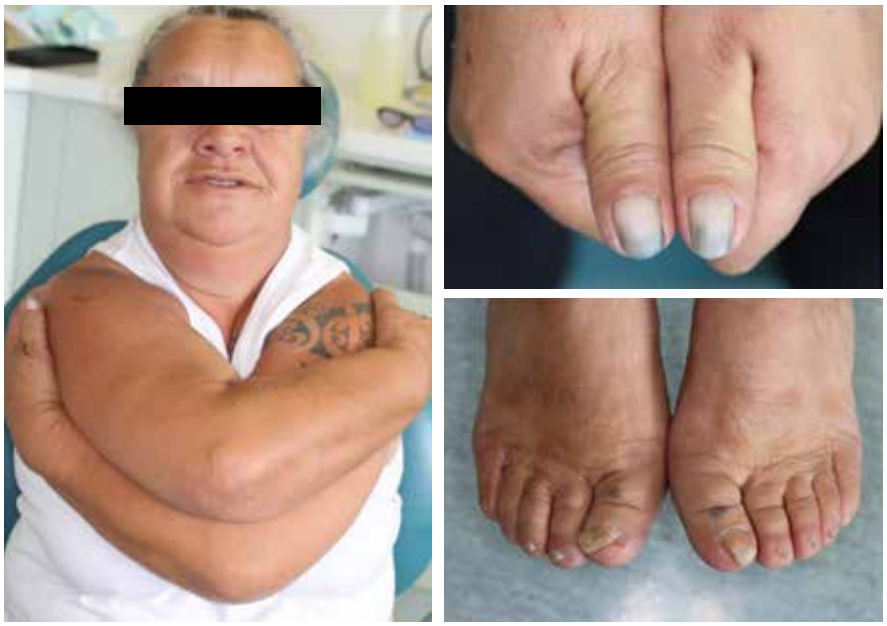

Figure 2. 3. 4: Clavicular aplasia, hands and feet.

Daskalogiannakis and co-workers (2006) involves a combination of orthodontic and maxillofacial surgery. In brief, these protocols involve timely extraction of deciduous teeth, staged surgical removal of supernumerary teeth, the exposure of selected permanent teeth and orthodontic forced eruption. Roberts *et al.* (2013) report that these procedures are all undertaken over a long period of time; patient compliance is essential for a favourable outcome.

The aim of dental management in CCD is to achieve an optimal functional and cosmetic result by early adulthood, so a multidisciplinary approach is necessary. Depending on the type and severity of the anomalies present, a team of maxillofacial surgeons, orthodontists and prosthodontists may be needed to develop an individualized treatment protocol (Roberts *et al.*, 2013) The commitment of the patient is crucial. The literature shows that the dental management of CCD has undergone a metamorphosis from a "wait and observe" approach to more sophisticated and costly methods combining orthodontics and surgery. Although there are numerous options, there is a general consensus that the best results are obtained if the condition is diagnosed and treated early.

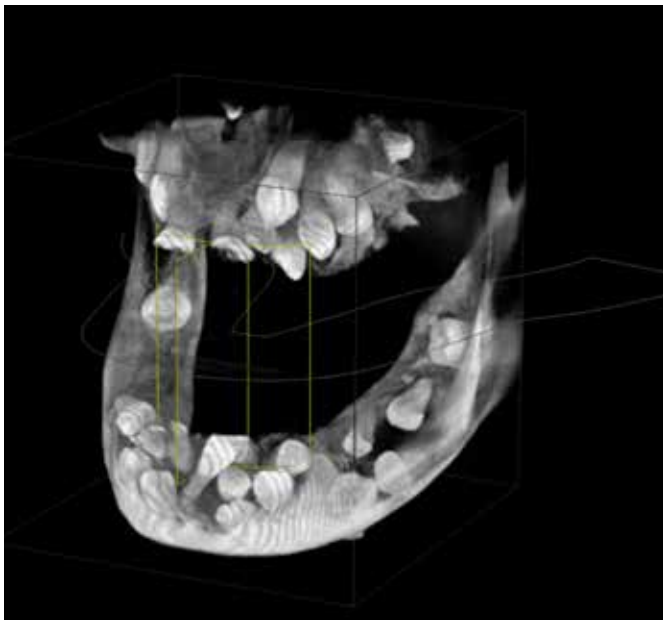

Figure 5: Cone beam CT

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