

# Case of odontoma-related infection in a cleidocranial dysplasia

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## DESCRIPTION

Cleidocranial dysplasia (CCD) is a rare syndrome with an estimated prevalence of 1:1 000 000.<sup>1,2</sup> It has an autosomal dominant transmission with complete penetrance and variable expression, equally affecting men and women.<sup>3</sup> This skeletal disorder is caused by a mutation in the *RUNX2* (CBFA1) gene, on the short arm of chromosome 6 (6p21).<sup>3</sup> This gene encodes a runt-related transcription factor involved in osteoblast and chondrocyte differentiation.<sup>3</sup> The most frequent clinical features are skull deformity, hypoplastic maxilla, hypoplastic or aplastic clavicles, supernumerary teeth and delayed teeth eruption.<sup>3</sup> Although these features may vary, normal deciduous dentition, delayed eruption of permanent teeth and multiple supernumerary teeth impaction are practically pathognomonic of CCD.<sup>3</sup>

Supernumerary teeth and odontomas (ODs)—the most frequent odontogenic benign tumours—share a common origin.<sup>4</sup> They arise from a hyperactive dental lamina as a response to teratogenic or genetic stimuli.<sup>4</sup> Mostly asymptomatic, both can be related with delayed tooth eruption.<sup>4</sup> Composed of enamel, dentine and cement, ODs can be divided into compound and complex.<sup>5</sup> While compound ODs consist of multiple little tooth-like structures usually in the anterior upper maxilla, complex ODs are single amorphous masses without structural organisation, generally found in the posterior mandible.<sup>5</sup>

Throughout the years, different ways of dental management were proposed, namely the Toronto–Melbourne, the Belfast–Hamburg, the Jerusalem and the Bronx approaches.<sup>3</sup>

Besides having small differences in the timing and number of interventions, all of these approaches defend the extraction of deciduous



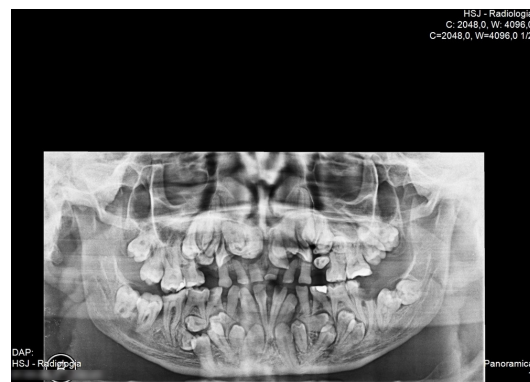
**Figure 2** Axial CT demonstrating a well-defined radiopaque mass, surrounded by a radiolucent lesion in the right canine incisor area. Multiple supernumerary teeth.

and supernumerary teeth and orthodontic treatment afterwards.<sup>6</sup> Several cases need extensive and complex prosthetic rehabilitation.<sup>6,7</sup>

Because of the complexity of these cases, the management is multidisciplinary and case-by-case oriented.<sup>7</sup> We did not find any evidence of an approach demonstrating higher rates of success when compared with others.



**Figure 3** Intraoral X-ray showing the radiolucent lesion extending to the apex of 5.1.

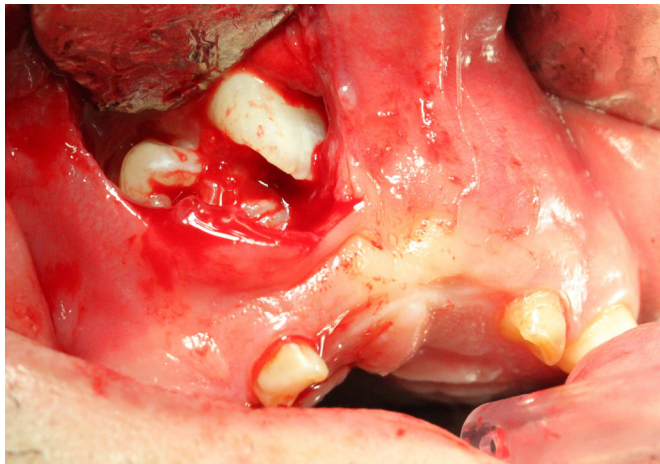


**Figure 1** Orthopantomography showing multiple impacted teeth, including supernumerary ones, and a radiopaque mass in the first quadrant.



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**Figure 4** View of maxillary right central incisor (right) and odontoma (left).

A 26-year-old man was referred to our department due to repeated and refractory dental infections and multiple impacted teeth. At time of the appointment, he was under treatment with a beta-lactam antibiotic.

The patient had been diagnosed with CCD since early childhood but, apparently, without dental management. Besides CCD, the patient had no other known conditions. Familywise, there are three more diagnosed cases of CCD (the mother and two siblings).

Intraoral examination showed the presence of early mixed dentition, severe pain on percussion of the tooth 51 and multiple carious lesions.

Panoramic X-ray (figure 1) and CT scan (figure 2) revealed multiple impacted supernumerary teeth and a well limited radiopaque mass, with a radiolucent halo, in the right canine incisor area.

After an intraoral X-ray (figure 3) showing the radiolucent halo extending to the apex of 51 and due to the refractory



**Figure 5** Complex odontoma (left) and right central incisor (right).

symptoms, this tooth was extracted with purulent discharge. The curettage of the dental alveolus provided enough material to histopathological examination that showed multiple fragments of epithelium with areas of inflammatory infiltrate compatible with an inflammatory cyst.

Because of the anxiety showed by the patient during oral examination and the uncooperative behaviour for medical treatments reported by him and his mother, he underwent general anaesthesia to extract 52, impacted 11 and a complex OD (figures 4 and 5). Other dental procedures were performed due to the pre-existing carious lesions.

Eight months later, the patient remains asymptomatic.

As previously mentioned, the hyperactivity of the dental lamina promoted the development of supernumerary teeth.<sup>4</sup> While supernumerary teeth and dental malformations are expected in CCD, the existence of ODs and OD-related infections are less common.<sup>5 8 9</sup>

Therefore, this is a typical case of CCD associated with an uncommon OD (complex) in an unusual location (maxilla).

### Learning points

- ▶ Cleidocranial dysplasia (CCD) is a rare disease, with no universally accepted protocol; hence, it is desirable that the patient should be referred to a multidisciplinary team.
- ▶ Dental management should occur since early childhood.
- ▶ While supernumerary teeth and dental malformations are expected in CCD, the existence of odontomas (ODs) and OD-related infections are less common.

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