Cleidocranial dysplasia: a case report

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ABSTRACT

Cleidocranial dysplasia is a rare, autosomal dominant congenital disorder. A 12-year old female visited with chief complaint of unerupted permanent teeth. Also her father showed severe class III malocclusion. The extraoral radiography and computed tomography showed delayed closure of the cranial sutures and underdevelopment of maxilla, maxillary sinuses, and frontal sinus. Both clavicles were underdeveloped and thoracic rib cage was bell-shaped. Both zygomatic process appeared as hypoplastic feature. There were many unerupted permanent and supernumerary teeth in the maxilla and mandible. We examined location and number of the unerupted teeth using 3D CT. Finally we could conclude this case was cleidocranial dysplasia based on the clinico-radiologic findings. (***Korean J Oral Maxillofac Radiol 2005; 35 : 225-9***)

KEY WORD : Cleidocranial Dysplasia; Tomography, X-ray Computed

Cleidocranial dysplasia is a rare congenital disorder and an autosomal dominant skeletal dysplasia.[1,2] This disease is characterized by abnormality of the clavicles, skull, and other part of the skeleton, together with a tendency toward retention of the primary dentition, failure of the permanent teeth to erupt, and impacted supernumerary teeth.[1,3-5]

Cleidocranial dysplasia is associated with mutation of runt-related gene 2 (Runx2), also referred to as Cbfa1 on chromosome 6p21, a transcriptional factor essential for osteoblast and dental cell differentiation as well as bone and tooth formation. Recently, studies have demonstrated that Runx2 gene is involved in fetal and postnatal growth of bone and postnatal formation of dental tissue and tooth eruption.[6-10] Both dominant and recessive pattern of inheritance have been described. It is known that spontaneous mutation occur in 20-40% of cleidocranial dysplasia patients.[11]

This dysplasia was first described by Marie and Sainton in 1898.[5,12-15] Marie and Sainton coined the descriptive title cleidocranial dysostosis. The disorder was firstly thought to involve bones of intramembranous origin only, but it is now known that bones of endochondral ossification are also affected. So the cleidocranial dysplasia was substituted for cleidocranial dysostosis to show the more generalized nature of the condition.[4,16]

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Skeletal complications are common in this disease. There are rudimentary or completely absent of clavicles resulting in the ability of the patient to approximate their shoulders, vertebral abnormalities, thoracic deformities with narrow thorax and hand and feet abnormalities.[1-5,17]

The skull and facial complications may also be common. Delayed ossification of the cranial suture and fontanelles occur. As a result of the abnormal ossification, wormian bones can be seen. There are also hypoplastic maxillary and nasal bone, very small or absent maxillary sinuses, and small anterior nasal spine often directed in an inferior direction.[4,5,9] The most striking oral abnormalities are the failure of eruption of the permanent teeth with multiple unerupted supernumerary teeth.[4,5]

These deformities may lead to the characteristic appearance of short stature, narrow, and drooping shoulders, bossing of frontal and parietal bones, small face in relation to the cranium, and ocular hypertelorism.[4]

Jensen and Kreiborg have in recent years provided reports that the ascending ramus was parallel-sided and that the coronoid pointed with a distal curvature in cleidocranial dysplasia patients. They also reported that in cleidocranial dysplasia the zygomatic bones were hypoplastic, thin or even discontinuous at the zygomaticotemporal suture, ranging in width from 1 mm to 5 mm.[18]

Mcnamara et al. reported summary of the feature of cleidocranial dysplasia on panoramic radiography and 9 critical features. They also concluded dental panoramic radiography

is a valuable adjunct in confirming the diagnosis of cleidocranial dysplasia.[4]

Recently we have examined the patient that showed characterized feature of eruption failure of the permanent teeth with multiple unerupted supernumerary teeth, and other skeletal abnormalities. We present a case of cleidocranial dysplasia examined by 3D CT for the localization of multiple unerupted permanent and supernumerary teeth.

Case report

A 12-year old female visited Kangnung National University Dental Hospital with chief complaint of uneruption of permanent teeth. Previous medical history was normal. She treated in local dentistry for extraction of some prolongedly retained primary teeth.

This patient showed the appearance of ocular hypertelorism and depressed nasal bridge (Fig. 1). The intraoral examination showed a Class III malocclusion with the permanent first molar in crossbite, prolongedly retained primary teeth, and crossbite in anterior primary dentition (Fig. 2). The intraoral clinical examination showed primary dentition except for four first molars and two lower central incisors. Also her father showed severe class III malocclusion.

On panoramic radiographic examination, we could find the eruption failure of multiple permanent teeth with multiple

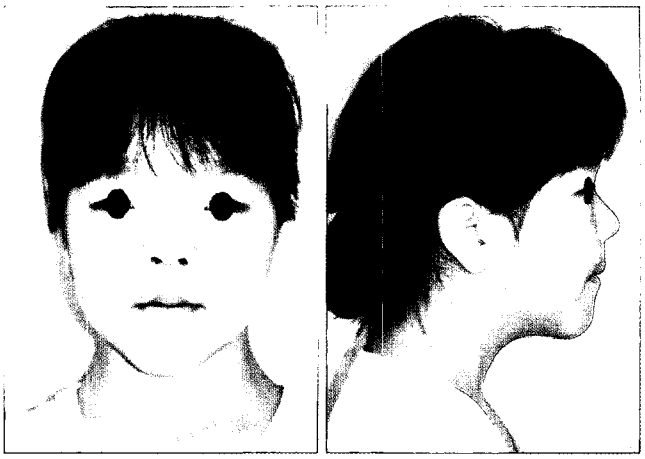

Fig. 1. Extraoral finding shows the appearance of ocular hypertelorism and depressed nasal bridge.

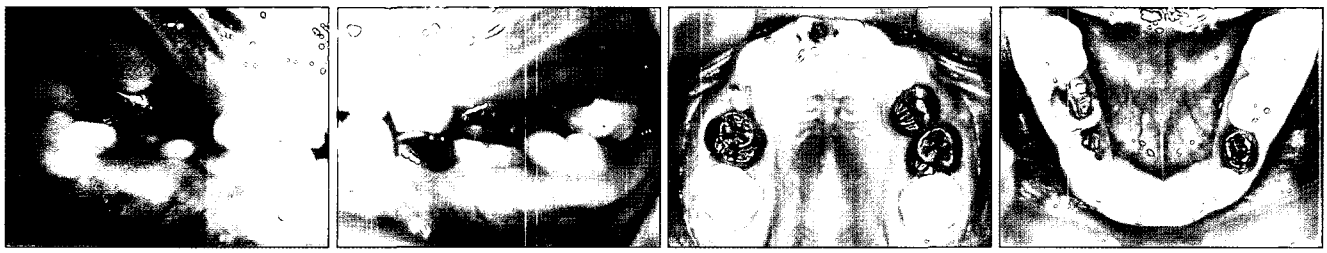

Fig. 2. The intraoral clinical examination shows primary dentition except for four first molars and two lower central incisors. The permanent first molar shows Class III malocclusion.

Fig. 3. Panoramic radiograph shows the eruption failure of multiple permanent teeth with multiple unerupted supernumerary teeth in both jaws. The ascending ramus is parallel-sided on both sides.

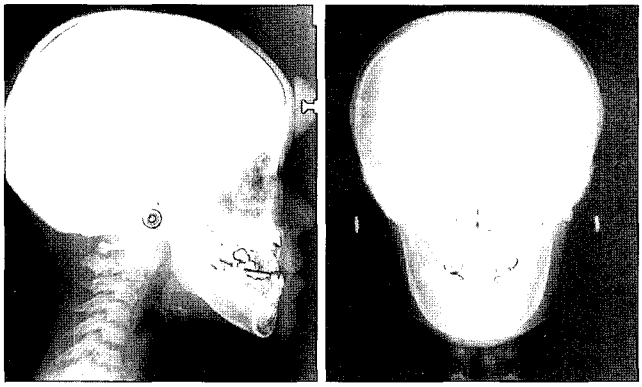

Fig. 4. Lateral and postero-anterior cephalometric radiographs show unclosed coronal, lambdoid and sagittal suture, and multiple wormian bone in the rambdoid suture on skull. Zygomatic bones are discontinuous at the zygomaticotemporal suture. Maxillary hypoplasia is mild.

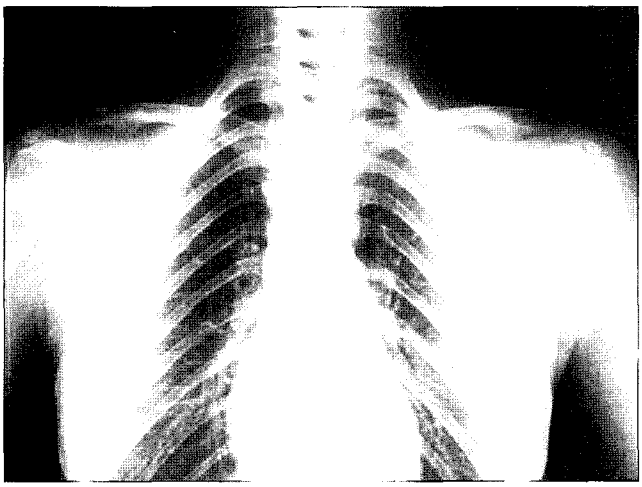

Fig. 5. Chest radiograph shows slightly wide space between clavicle and acromion on both sides but hypoplasia of clavicles are slight. Thorax is narrow and bell-shaped.

unerupted supernumerary teeth in both jaws. There were about 13 supernumerary teeth. We could also find that the ascending ramus was parallel-sided on both sides (Fig. 3).

On lateral and postero-anterior cephalometric radiographs, we could find unclosed coronal, lambdoid and sagittal suture, and multiple wormian bone in the rambdoid suture on skull. Zygomatic bones were discontinuous at the zygomaticotemporal suture. Maxillary hypoplasia was mild and even SNA value was normal (Fig. 4).

On chest radiograph, we could find slightly wide space between clavicle and acromion on both sides but hypoplasia of clavicles were slight. Thoracic deformities were also shown with narrow and bell-shaped thorax (Fig. 5).

On reformatted 3D CT images of teeth, we found an additional supernumerary tooth uncounted on panoramic view. This tooth located on left maxillary area (Fig. 6)

On the basis of CT image, craniofacial model of this patient could be made. From this model we could confirm discontinuity of zygomatic bone and found hypoplastic nasal bone.

We could diagnose this case as a cleidocranial dysplasia from clinical and radiological examinations. Treatment began with the removal of primary teeth, impacted supernumerary teeth and surgical exposure with attachment of brackets on unerupted permanent teeth for additional orthodontically guided eruption. And remaining treatment procedures are in progress within the framework of a multidisciplinary approach.

Discussion

Cleidocranial dysplasia is an rare disease with a prevalence of less than 1 per million.[2] More than 100 other abnormalities have also been associated with clinical features of this condition.[13] The clinical variability of cleidocranial dysplasia

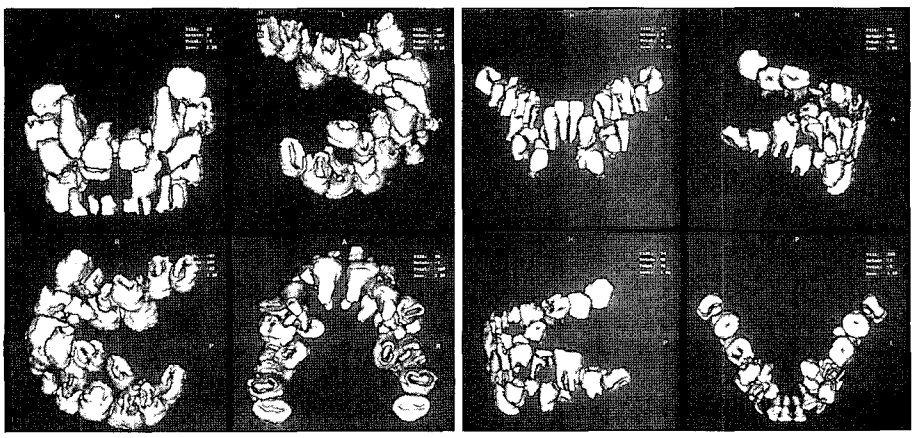

Fig. 6. Reformatted 3D CT images of teeth shows supernumerary teeth in both jaws.

ranges from nearly unrecognizable to full-blown case. Because of the relative lack of medical complication, it can be underdiagnosed.[2]

Characteristic features include clavicular deformities, thoracic deformities, abnormalities of dentition, patent fontanelles, wormian bone, and a variety of other minor skeletal abnormalities.[1-5,18] The clavicles are frequently of defective development, and they may rarely be completely absent, although it is more common for some part of both to form, the outer end of one and the inner end of the opposite one, or the same portions of each. Most case of cleidocranial dysplasia have some developmental abnormality of the dentition, slight or gross. The primary dentition is usually normal, although may be retained in the arch throughout life. It is common for permanent teeth not to erupt above the gum, or to be only partially erupted. Indeed, the presence of several well-formed supernumerary teeth is highly suggestive cleidocranial dysplasia. The cranial fontanelles are very slow to close and may never close, even late in life. Extra sutures are frequently present with multiple wormian bones, most of which are seen in the occipital bone.

The maxilla is sometimes small and the mandible is usually of normal size. As a consequence there may be some prognathism. But, in the other study, Jarvinen reported real maxillary prominence in addition mandibular prognathism in cleidocranial dysplasia patient as an atypical case.[19]

In our case, we could see slightly wide space between clavicle and acromion on both sides but hypoplasia of clavicles were slight. And thorax was slightly narrow and bell-shaped. The patient were characterized by abnormalities of dentition in addition to wormian bone. We could find the features of typical cleidocranial dysplasia in the primary and permanent dentition. We could find 14 supernumerary teeth in the maxilla and mandible. In cephalometric view, we could find wide lambdoid sutures and suture with wormian bone in occipital area. On the other hand, the patient had atypical features of cleidocranial dysplasia-relatively normal maxilla by cephalometric analysis. Golan et al. concluded that a concave facial profile was a result of a hypoplastic midface and a prognathic mandible influenced by the pubertal growth spurt, and should therefore only be used as indicators in adults.[17] So we could think our case as an individual variation of the cleidocranial dysplasia on some aspects.

Jensen and Kreiborg have provided reports that the ascending ramus was parallel-sided and that the coronoid pointed with a distal curvature in cleidocranial dysplasia patients. In addition, they reported that there may be a thickening of the ascending ramus on the lingual side between the inferior dental canal and the internal oblique ridge. They also confirmed that in cleidocranial dysplasia the zygomatic bones are hypoplastic, the maxillary sinuses may be absent or very small, and the nasal bones are hypoplastic.[18] Comparing these features with our case, We could find that all these radiologic features excluding the thickening of the ascending ramus, can be identified on a panoramic radiograph.

We used reformatted 3D CT images of teeth and craniofacial model on the basis of CT image as a supplementary diagnostic tool. Using reformatted 3D CT images of teeth, we found an additional supernumerary tooth that we had not detected on panoramic radiography. It is suggested that reformatted 3D CT images of teeth should be valuable diagnostic tool in cleidocranial dysplasia patient especially when there are numerous supernumerary teeth superimposing with the permanent teeth.

Treatment of cleidocranial dysplasia usually begins with the removal of primary teeth and supernumerary teeth to help permanent teeth erupt. Becker et al. presented an orthodontic and surgical method, the Jerusalem approach that involves 2 stages of surgically uncovering and bonding the permanent teeth. The first stage is to guide the incisors into occlusion, and the second stage is to guide the remaining permanent teeth into occlusion.[20] Cooper et al. concluded that management of dental abnormalities is best accomplished by a team approach with long-term follow up of specialists who are familiar with the growth and development.[2]

In our case, we removed primary, and impacted supernumerary teeth and exposed unerupted permanent teeth with attachment of brackets for additional orthodontically guided eruption. And remaining treatment procedures are in progress within the framework of a multidisciplinary approach.

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