CASE REPORT

Cleidocranial Dysplasia in a 10-year-old Child: A Case Report

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Abstract

Cleidocranial dysplasia is a rare congenital anomaly characterized by multiple skeletal defects of which partial or complete absence of clavicles, delayed closure of fontanels with presence of open sutures and multiple wornian bones forms a striking feature. The oral manifestations are delayed exfoliation, delayed or failing eruption of the permanent dentition with multiple supernumerary teeth, protruding mandible and mid face retrusion. It is also known as Marie and Sainton's disease, mutational dystosis or cleidocranial dysostosis. A 10-year-old female patient reported to the Department of Pedodontics and Preventive Dentistry, Sathyabama University, Dental College and General Hospital, Chennai, Tamil Nadu with the chief complaint of unerupted permanent teeth. Radiological investigations confirmed the diagnosis of cleido cranial dysplasia and various treatment options are discussed in this article.

Keywords: Cleidocranial dysplasia, Fontanels, Supernumerary teeth.

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Background

Cleidocranial dysplasia is a rare congenital anomaly characterized by multiple skeletal defects, of which partial or complete absence of clavicles, delayed closure of fontanels with the presence of open sutures, and multiple wornian bones form striking features.[1] The oral manifestations include delayed exfoliation, delayed or failing eruption of the permanent dentition with multiple supernumerary teeth, protruding mandible, and mid-face retrusion.[2] It is also known as Marie and Sainton's disease, mutational dystosis, or cleidocranial dysostosis. Patients with CCD are likely to have a high-arched or cleft palate, short stature, and scoliosis of spine with short stature.[3]

Case Description

A 10-year-old female patient reported to the Department of Pedodontics and Preventive Dentistry, Sathyabama University Dental College and General Hospital, Chennai, Tamil Nadu, with the chief complaint of unerupted permanent teeth.

On general examination, the patient was of short stature, and the weight was normal to her age. Further examination revealed a brachycephalic skull, frontal bossing, hypertelorism, and sloping of shoulders (Fig. 1). The facial symmetry was normal with deficit in midface, concave profile, and competent lips (Fig. 2).

Intraoral examination revealed that the child had retained deciduous teeth along with lower first permanent molars with delayed eruption of the permanent teeth. Root stumps were seen

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Fig. 1: Frontal view of the child's physical stature

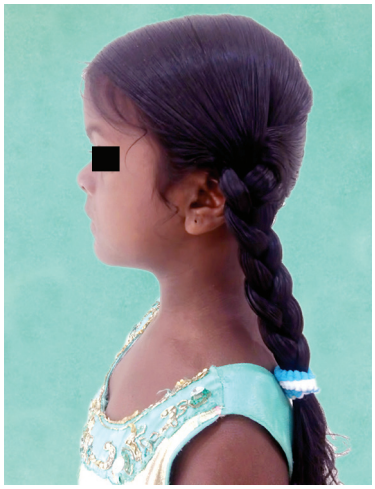

Fig. 2: Lateral profile view exhibiting midface deficit and concave profile

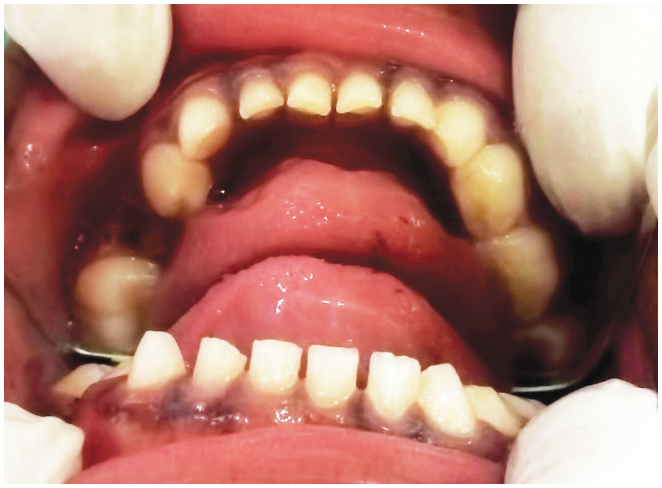

Fig. 3: Intraoral view of mandible with multiple retained deciduous teeth and root stumps in relation to 84

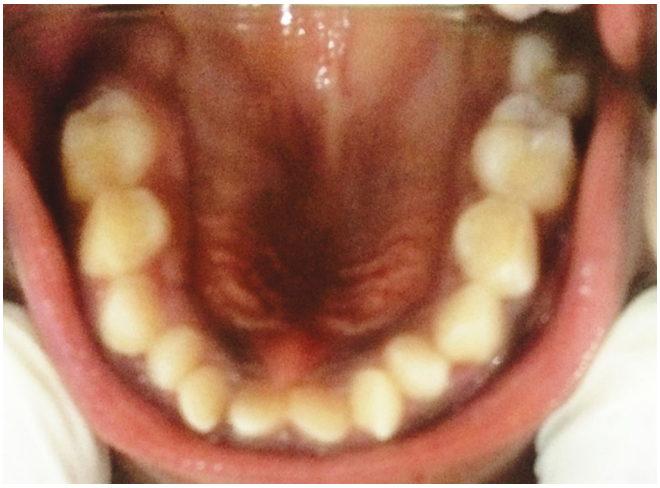

Fig. 4: Intaoral view of maxilla with multiple retained deciduous teeth and high arched palate

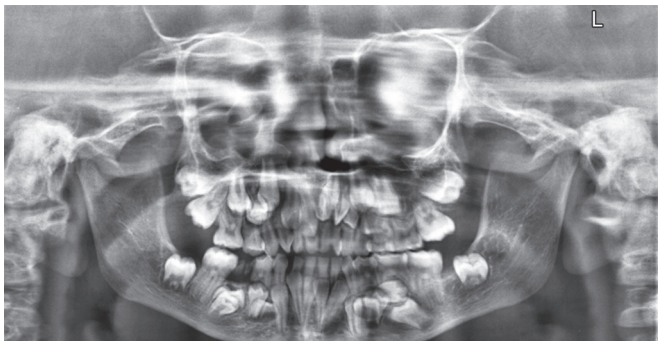

Fig. 5: Panoramic view of the child with multiple supernumerary teeth in premolar region

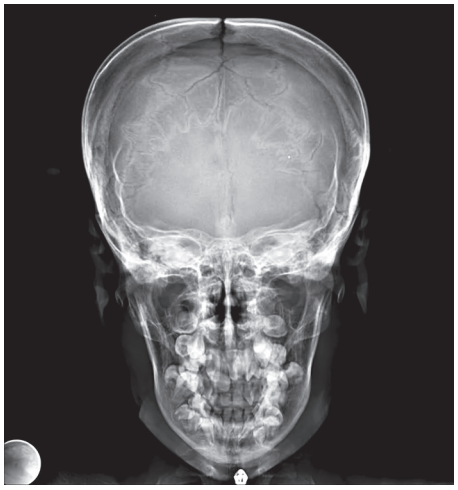

Fig. 6: Anteroposterior view skull CT with patent anterior fontanel

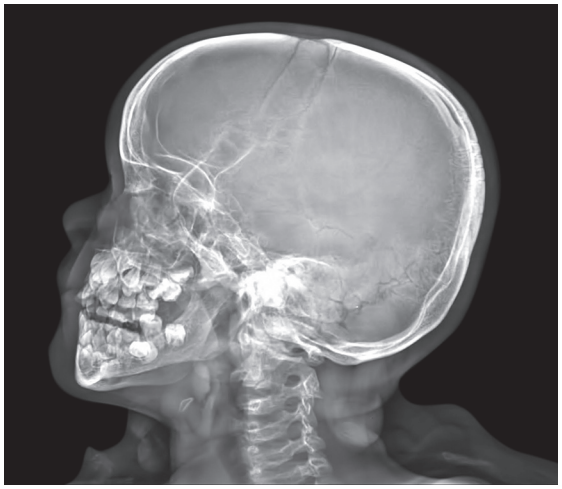

Fig. 7: CT (lateral view) of the skull with open fontanels and delayed closure of sutures

in relation to 85 (Fig. 3). On further examination, it was noticed that there was a prominent maxillary retrusion with a high-arched palate (Fig. 4).

Extraction of the root stump with relation to 85 had been planned for the child. Radiographical investigations were planned prior to the extraction to confirm the presence of underlying permanent tooth buds. Panoramic radiograph (OPG) revealed multiple impacted supernumerary teeth in the upper and lower premolar regions with retained deciduous teeth, coinciding with the delayed eruption of permanent teeth (Fig. 5).

Furthermore, a CT skull in AP and lateral views and a PA chest X-ray were advised to rule out any skeletal abnormality. The AP view of the skull depicts the presence of widened sutures and open anterior fontanels with the presence of multiple wornian bones (Fig. 6). Also, the lateral view depicts the presence of delayed ossification of saggital sutures along with a decreased anteroposterior length of the maxilla when compared to the mandible (Fig. 7). In addition, a PA view of the chest showed a hypoplasia of clavicular bones with a funnel-shaped chest, which confirmed the diagnosis of cleidocranial dysplasia (CCD) as was further confirmed by clinical findings (Fig. 8).

Discussion

Cleiodocranial dysplasia is a rare syndrome usually having autosomal dominant inheritance.[4] It is known that CCD is caused by heterozygous mutations in RUNX2 gene, which encodes a transcription factor required for osteoblast differentiation and is located on chromosome 6p21.[5]

The main defect of this disorder lies in faulty ossification of selected endochondral and intramembranous bones. This is an

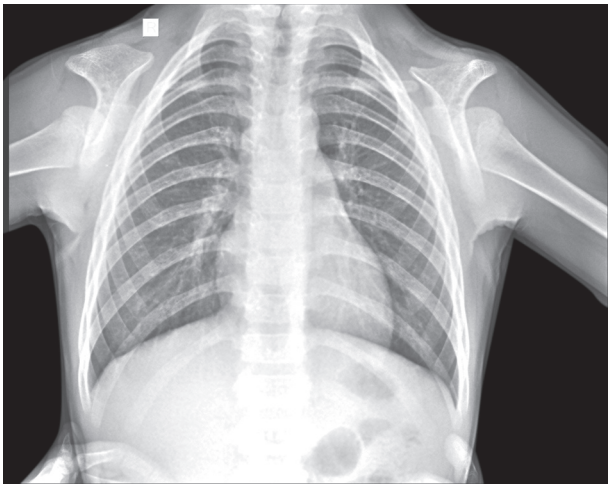

Fig. 8: Posteroanterior chest radiograph of the child exhibiting clavicular hypoplasia

Fig. 9: Parent's physical stature showing clinical features of the syndrome

Fig. 10: Lateral profile view showing midface deficit with retrusion

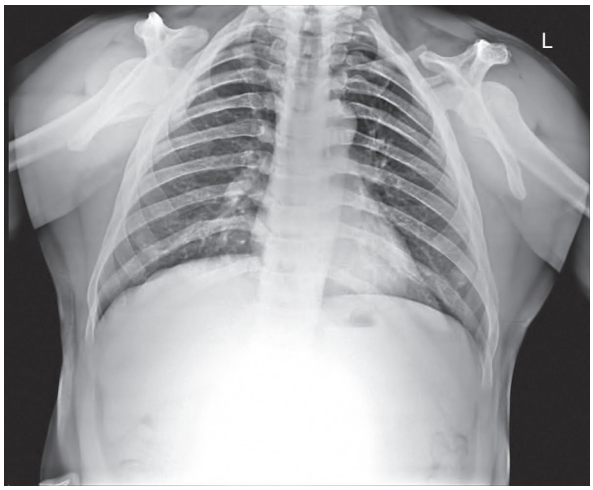

Fig. 11: Posteroanterior chest radiograph of child's father exhibiting clavicular hypoplasia

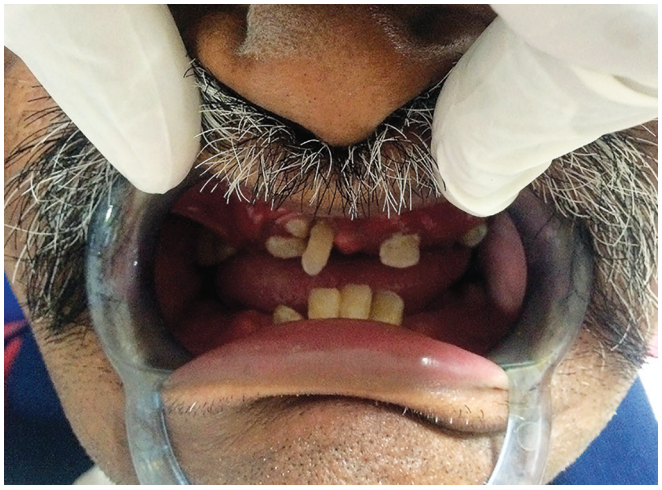

Fig. 12: Intraoral view of the child's parent with multiple supernumerary teeth

early developmental disorder of mesenchyme or connective tissue, producing retarded ossification of the membranous and cartilaginous precursors of the bone, especially at the junction of various bones. This may lead to delayed or even failure of ossification of portions of the skeletal structure. CCD involves the bones that are ossified earliest in fetal life, especially the clavicle. The disease classically causes a retardation or partial aplasia of bones that ossify intramembraneously.[5,6]

To confirm the genetic history, we checked the history of the parents and grandparents of the patient and there were positive findings that correlated with the father and the grandparents who had the same physical appearance and intraoral findings (Figs 9 to 12). A pedigree chart was done, which futher confirmed the autosomal dominant mode of inheritance of the disorder (Fig. 13).

Orofacial findings of this disorder are very significant. Extraoral findings include an increase in the transverse dimension of the cranium (brachycephalic head) and saggital shortening of the cranial base. Frontal and parietal bossing may be associated along with metopic depression. Closure of the fontanels and cranial sutures are delayed and wornian bones may be present (Figs 6 and 7).[7]

Priyanka et al. observed a similar case of cleidocranial dysplasia in a 5-year-old male child, with multiple impacted supernumerary teeth in lower premolar region born to nonconsanguineous couple.[8] Reddy et al. described that radiographs could be an important diagnostic aid for cleidocranial dysplasia.[9]

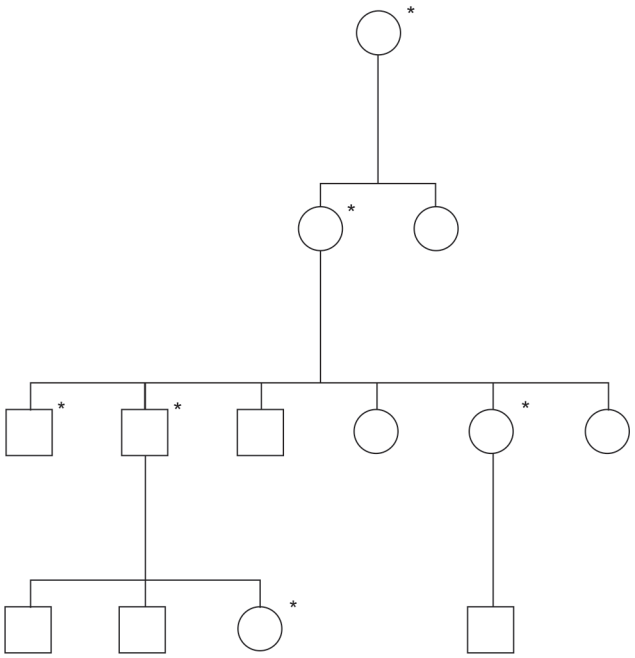

Fig. 13: Pedigree chart analysis
(*autosomal dominant mode of inheritance of the disorder)

McNamara et al. reported that dental abnormalities are typical main features of CCD in 93.5% of affected patients based on his observations on radiographs of multiple impacted permanent and supernumerary teeth in a 10-year-old male child.[10] Similar observations were seen in a case report by Sakhi et al.,[11] which correlated with our findings. Bhat et al. reported the presence of frontal and parietal bossing, open sutures, and fontanels with the presence of supernumerary teeth and malocclusion of teeth, which was also reported in our case.[4]

The treatment of cleidocranial dysplasia varies owing to the skeletal or dental abnormalities and the condition of impacted normal or supernumerary teeth. Dental management in cleidocranial dysplasia is challenging in most cases and aims to achieve functional and esthetic results.[12] Collaborative orthodontic-surgical treatment of dental and skeletal anomalies of the jaws is suggested. Several combined treatment regimens were proposed according to the timing and manner of approach to the impacted permanent teeth.[12]

The traction of impacted teeth with elastics is a common method used in contemporary orthodontics, especially in syndromic cases such as CCD. After surgical exposure of an impacted or partially impacted tooth, a button or bracket is bonded and elastics are applied between impacted and opposing permanent erupted teeth.[13] The dental management of CCD has undergone a metamorphosis from a "wait-and-observe" approach to more sophisticated and costly methods that combine orthodontics and surgery.[13] Tasar et al.[14] has advocated the extraction of all the primary and permanent teeth and placement with a removable prosthesis.

Conclusion

Cleidocranial dysplasia is an uncommon disorder; however, its clinical and radiological manifestations have been characterized. Early diagnosis allows proper orientation to the treatment and offers a better quality-of-life. A holistic approach takes care of all the aspects, including the primary pathology and psychological aspects. The dental management of CCD requires a multidisciplinary approach and indeed requires special skills from pediatric dentists to manage such cases.

Clinical Significance

This case report may provide many treatment options for CCD, as it is indeed a challenge to treat such pediatric cases to achieve proper and healthy smiles.

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