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CASE REPORT

CLEIDOCRANIAL DYSPLASIA: A CASE REPORT

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ABSTRACT

Cleidocranial dysplasia is a rare congenital disease. It is characterized by autosomal dominant inheritance pattern with equal sex distribution which is caused due to mutations in the Cbfa1 gene (Runx2) located on chromosome 6p21. It primarily affecting skull, jaws, teeth, clavicle along with other skeletal abnormalities. It presents with skeletal defects of several bones, like partial or complete absence of clavicles, late closure of the fontanels, presence of open skull sutures and multiple wormian bones. This rare syndrome is of utmost importance in dentistry due to presence of multiple supernumerary teeth, facial bones deformities and deranged eruption patterns .We are reporting a classical case of cleidocranial dysplasia in 12 year old patient.

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INTRODUCTION

The cleidocranial dysplasia, also known as Marie and Sainton Disease, Scheuthauer Marie-Sainton Syndrome and Mutational dysostosis (Shafer *et al.,* 1979) is a rare disease which can occur either spontaneously or by a dominant autosomal inheritance, with no predilection of genre or ethnic group (Silva *et al.,* 1995; Neville *et al.,* 2004) (article 2). The phenotype is characterized by general dysplastic bone formation manifested in typical malformations in the skull, the pelvis and the thoracic region (artical8) This disorder primarily affects bones showing intra-membranous ossification, i.e. calvarial bones and clavicles. Excessive mobility of the shoulder girdle is notedas clavicles are underdeveloped to varying degrees and are completely absent in approximately 10percent of cases.. Bell shaped small thoracic cage is noted having short ribs (article 9). These individuals are usually short. They have persistent fonticulus of the cranium or late closure of the same. The sutures can also remain opened, and the sagital suture presents itself depressed, giving the cranium a flat appearance. The parietal bones, frontal and occipital are proeminent, the paranasal sinus underdeveloped and many other cranial abnormalities might be present (Article 2).

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Characteristically, patients with cleidocranialdysplasia, show prolonged retention of deciduous dentition and delayed eruption of permanent teeth. Adults with cleidocranial dysplasia have mixed dentition in their oral cavities. Maxilla is also underdeveloped along with ill-formed paranasal sinuses. This condition is of clinical significance to every dentist due to the involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth (Rajeev Kumargarg, 2008).

CASE REPORT

A 12 year-old boy reported to Department of oral medicine and radiology he presented with his mother with the chief complaint of delayed eruption of permanent teeth. The was born healthy parents, and the family history was unremarkable. There was no significant past medical and dental history. The patient had normal gait and posture. He had normal intelligence and well oriented to surroundings. His vitals were normal. There was no sign of pallor, cyanosis and lymphadenopathy noted. He was thin, poorly built and short stature. On extra-oral examination brachycephalic head, frontal bossing, hypertelorism, underdeveloped maxilla, depressed nasal bridge was noted with concave facial profile with competent lips. He had shrugged shoulders with more than normal mobility of the shoulder girdle i.e. shoulder could be brought to the midline of chest. Intraoral examination revealed presence of the following teeth in the oral cavity,

6 C B A| A B C D E 6
6 C B A | A B C D 6

root stumps of E | D

E |D E Were seen

Reveals multiple over retained deciduous, missing permanent upper and lower anterior tooth and ankyloglossia seen.

There was class III malocclusion with underdeveloped maxilla and prognathic mandible. On the basis of clinical findings a provisional diagnosis of cleidocranial dysplasia has been suggested. The patient is advised for radiological investigations by panoramic radiograph, PA skull, PA chest, Lateral cephalogram, hand-wrist radiograph, the panoramic radiograph shows multiple impacted teeth along with supernumerary teeth and rounded gonial angles.

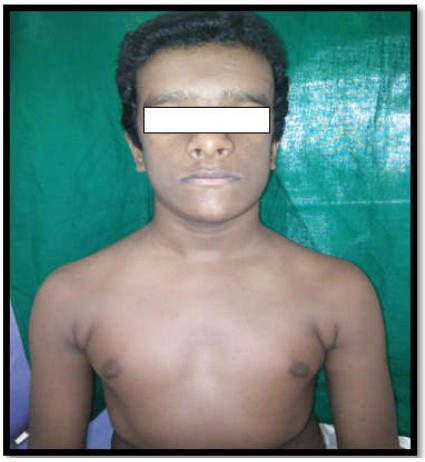

Figure 1. Extra-oral photograph showingprominent forehead, hypertelorismdepressed nasal bridge

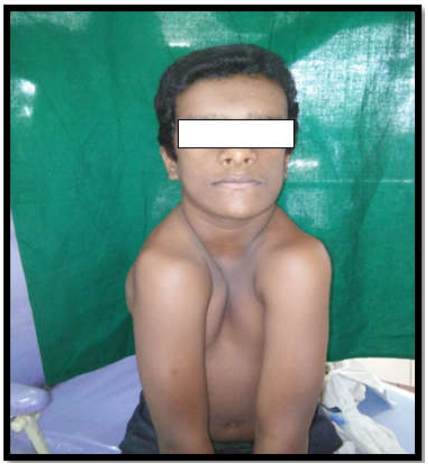

Figure 2. Frontal view of patient showingshrugged shoulders with more than normal Mobility of the shoulder girdle i.e. shouldercould be brought to the midline of chest

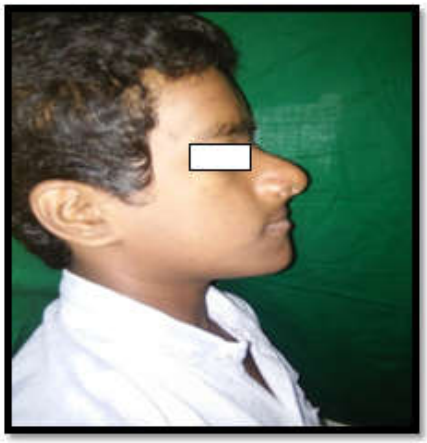

Figure 3. Patient's profile showing concavity due to underdeveloped maxilla

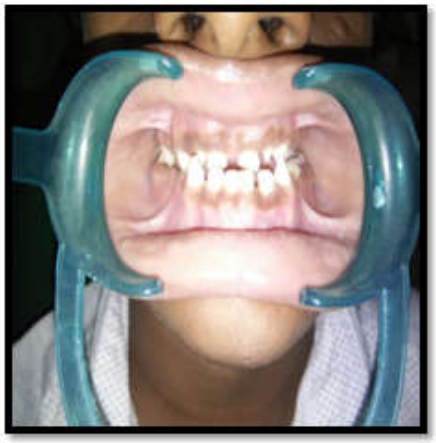

Figure 4. Intra-oral photograph showing all overetained deciduous teeth

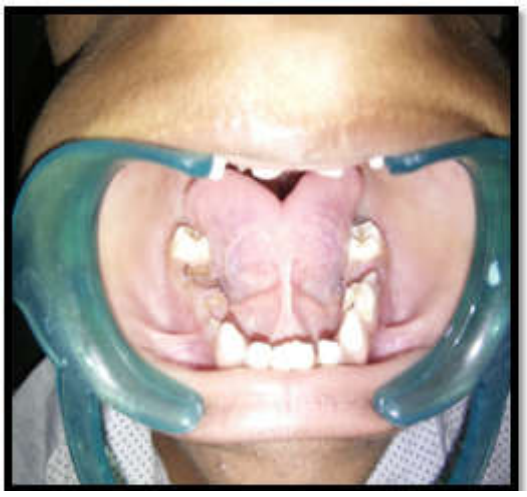

Figure 5. Intra-oral view showing Ankyloglossia and mandibular teeth

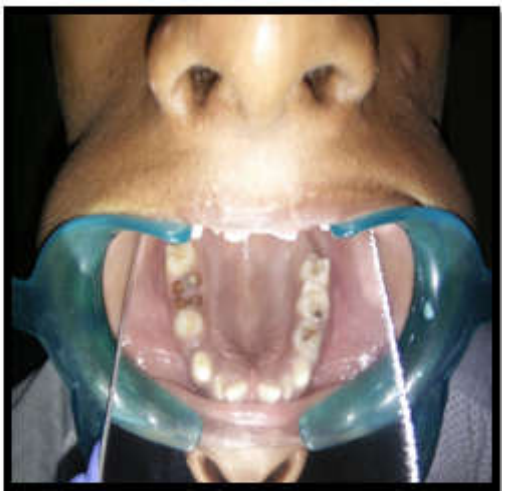

Figure 6. Intra-oral view showing maxillary teeth

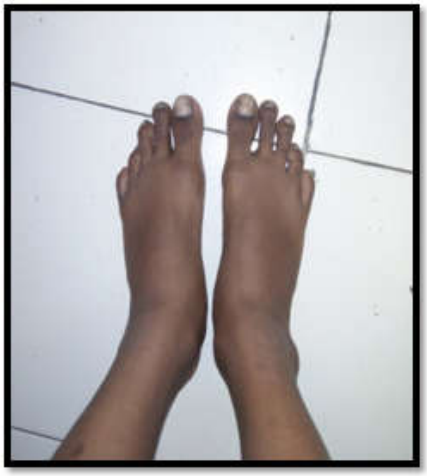

Figure 7 .

Figure 8.

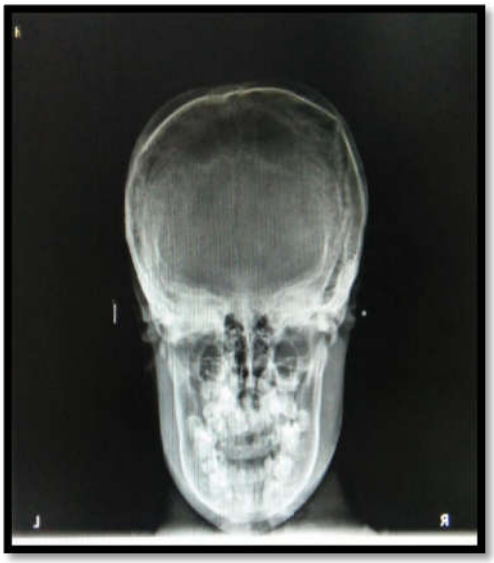

Figure 9. PA Mandible

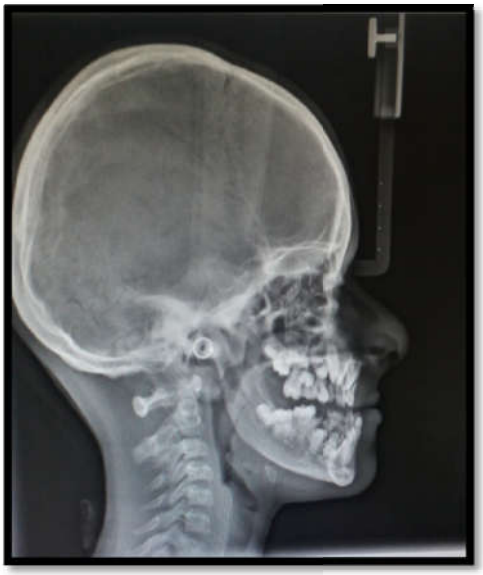

Figure 10. Lateral skull viewshowing wide open sutures with wormion bone

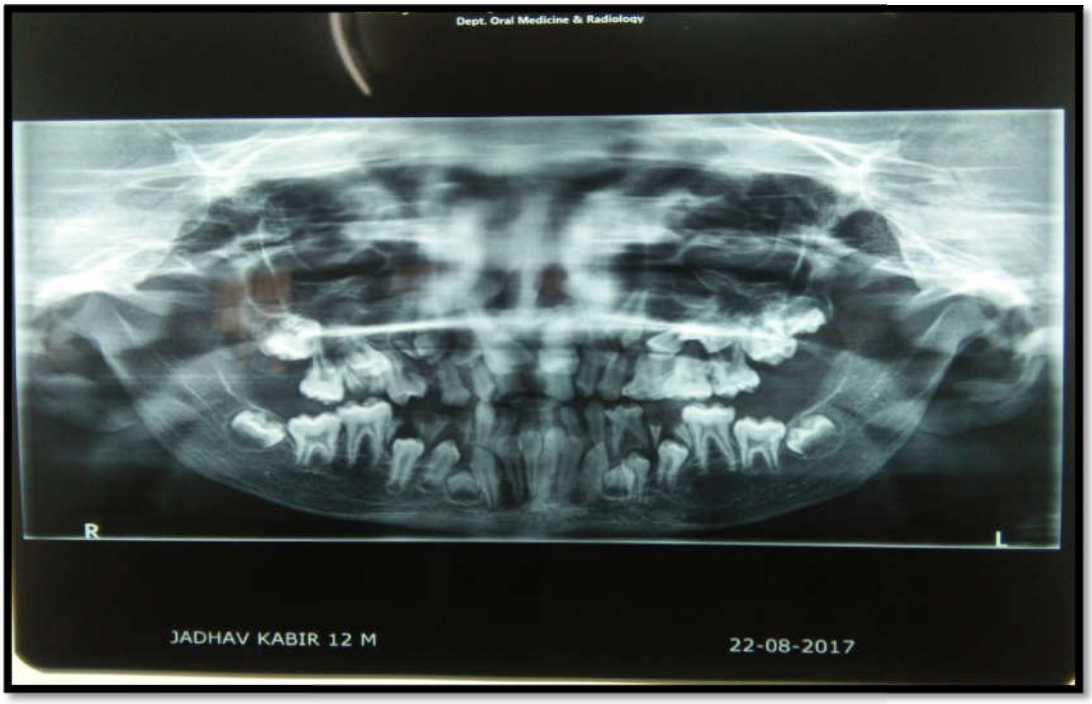

Figure 11. Orthopantomogram shows multiple impacted permanent and supernumerary teeth in bicuspid regions of the maxilla and mandible with over-retained deciduous teeth

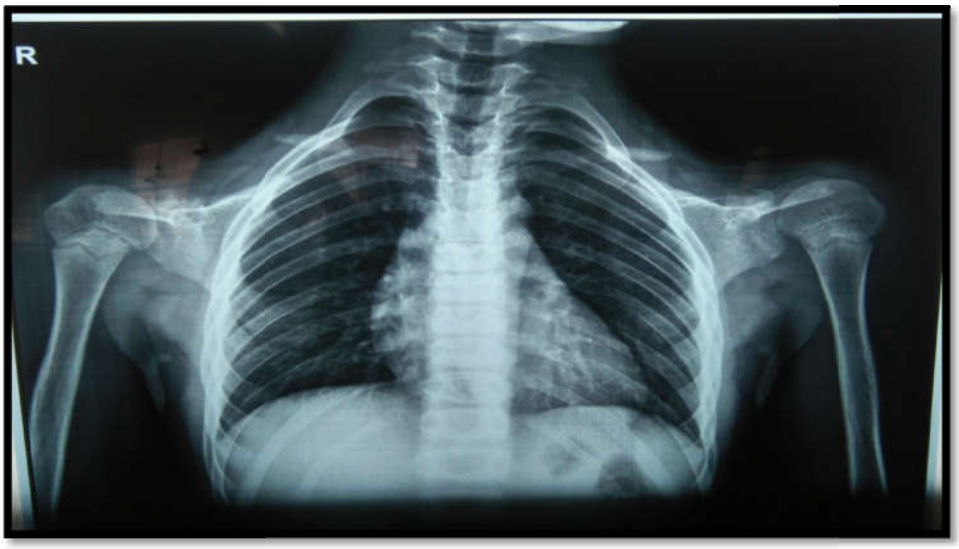

Figure 12. Chest X-ray

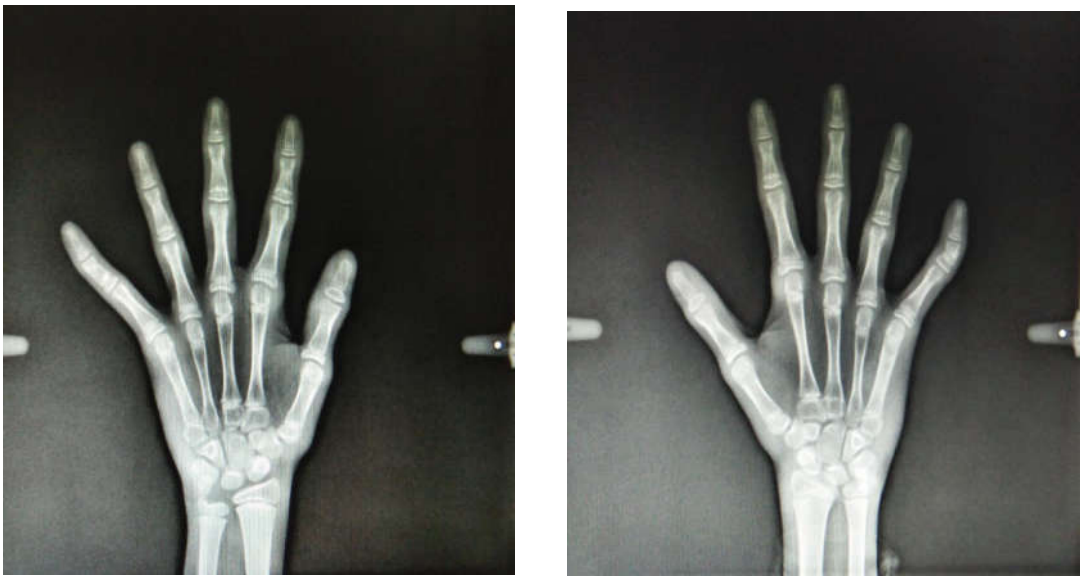

Figure 13. Hand wrist radiographs

PA skull shows widened anterior fontanel and posterior fontanels with presence of wormian bones, top of the metopic suture, sagittal suture and large mandible. PA chest radiograph shows thinning and hypoplasia of the clavicles and bell shaped rib-cage. Lateral cephalogram shows widened anterior and posterior fontanel with presence of wormian bones, nonfusion of saggital, coronal and lambdoid suture of skull bones and large mandible with impacted teeth and supernumerary teeth. Based on these clinical and radiological findings, the patient was diagnosed as a case of cleidocranial dysplasia.

DISCUSSION

The major features of Cleidocranial dysplasia are aplastic or hypoplastic clavicles, dental abnormalities (multiple supernumerary teeth, multiple impacted permanent teeth, retention of the deciduous teeth), and delayed closure of the sagittal fontanelles. Typically, our patient had all of these findings that are pathognomonic for a diagnosis of Cleidocranial dysplasia (Akhilan and Chaurasia, 2015). Most important and reliable tool to confirm the diagnosis is radiographic evaluation of patients. Broad sutures, large fontanels persisting into adulthood, numerous wormian bones and numerous unerupted supernumerary teeth are the pathognomonic radiological findings of Cleidocranial dysplasia. Vertebral defects with scoliosis, kyphosis or lordosis, pelvic bony abnormalities and anomalies of phalangeal, tarsal, metatarsal, carpal and metacarpal bones are also present. (SuhailRizvi and Hamid Raihan, 2006) Dental abnormality is one of the main features of CCD. Our patient had multiple supernumerary teeth, which can impede the normal eruption of permanent teeth. It has been suggested that supernumerary teeth in such cases should be removed as soon as possible (Anita Sharma *et al.,* 1995). 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. Many mutations in the RUNX2 gene have been identified in patients with CCD (Anita Sharma *et al.,* 1995). The differential diagnosis of cleidocranial dysplasia includes Apert syndrome, Dubowitz syndrome, Russell-silver syndrome, Down's syndrome and Crouzon syndrome 27 (Nivedita Ckvs *et al.,* 2016) the therapeutic approaches include:

- Prosthetic treatment. Following extraction of the impacted teeth or not the space in the dental arch is replaced with artificial teeth. In some cases, the

impacted teeth are exposed or dental implants are inserted to support the over dentures.

- Surgical treatment. Before surgical repositioning or transplantation of the permanent teeth, the supernumerary teeth should be removed.
- A combination of surgical and orthodontic treatment. After surgical removal of the deciduous teeth and imbedded supernumeraries, orthodontic treatment proceeds allowing eruption of the impacted permanent teeth and adjustment of the occlusion.

Conclusion

The clinical findings of cleidocranial dysplasia, although present at birth, are often either missed or diagnosed at a much later time. Some cases are diagnosed through incidental findings by physicians, treating patients for unrelated conditions. Cleidocranial dysplasia may be identified by family history, excessive mobility of shoulders and radiographic pathognomonic findings of the chest, skull and jaws (Rajeev Kumargarg, 2008).

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