Familial Occurrence of Cleidocranial Dysostosis: A Case Report

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ABSTRACT

Cleidocranial dysostosis is a rare congenital skeletal disorder, associated with total or partial absence of clavicles, delayed closure of cranial fontanelles and brachycephalic skull. There is delayed exfoliation of primary dentition and eruption of permanent teeth. Presence of multiple supernumerary teeth and morphologic abnormalities of the maxilla and mandible are also observed. The incidence of this autosomal dominant disorder is reported as 1:10,00,000 and the prevalence is 0.5:1,00,000, with no gender or ethnic group predilection. It was first described by Marie and Sainton in 1898. This paper presents the case of a seven year old male patient who reported to the department of Paediatric and Preventive Dentistry with a chief complaint of decayed teeth. Following thorough clinical and radiographic examination and investigations along with 3D images, the diagnosis was confirmed as cleidocranial dysostosis. A familial pattern was observed as patient's father and older sister had similar clinical features.

KEYWORDS: Cleidocranial dysostosis, congenital, impacted supernumerary teeth, hypoplastic clavicles, familial

INTRODUCTION

Cleidocranial dysostosis is a rare congenital hereditary disorder of autosomal dominant inheritance. It was first described by Pierre Marie and Paul Sainton in in 1898, hence is also known as Marie and Sainton disease, or Mutational dysostosis or Cleidocranial dysplasia.[1] The condition is usually caused by mutation in RUNX2 gene, necessary for functioning of osteoblasts.[2]

Cleidocranial dysostosis affects bones undergoing intramembranous ossification. It is characterized by partial or complete absence of clavicles causing hypermobility of shoulders, late closure of fontanelles, presence of open skull sutures, wide pubic symphysis and multiple wormian bones. Delayed closure of anterior fontanelle and metopic sutures causing frontal bossing is observed. Dysplastic skull base and reduced growth resulting in increased skull width leading to brachycephaly and hypertelorism is a typical feature.[1,2,3] Thoracic cage is small and bell shaped with short ribs.[1]

Characteristically patients with cleidocranial dysostosis show dental abnormalities like prolonged retention of deciduous dentition and delayed eruption of permanent teeth. Multiple supernumerary teeth and impacted teeth is a frequent finding. Crown and root abnormalities may be seen.[1,2,3]

This article aims to report the familial occurrence of this condition, involving the patient's father and sister.

CASE REPORT

A seven year old boy reported to the Department of Pedodontics & Preventive dentistry at Sinhgad Dental College & Hospital, Pune, with a chief complaint of decayed teeth. His medical history was non-contributory. He had undergone dental treatment in the past, with reported history of extraction of 54 & 65 due to caries and pulpectomy procedure for 75, 84 & 85. General physical examination revealed the patient to be thin built, of short stature, having a long neck, narrow thorax, narrow sloping shoulders with the ability to oppose them (Figure 1). Extraoral features revealed brachycephalic skull, frontal bossing with deep median furrow, parietal and occipital bossing and hyperteloric eyes (Figures 2,3). He presented with normal intelligence. On interviewing, and following general examination, the patient's father and older sister also presented with similar extraoral features (Figures 4a, 4b, 5a, 5b).

Intraoral examination of the patient revealed high arched palate, multiple carious deciduous teeth and restorations, with missing 54 and 65 (Figures 6,7). Retention of primary teeth and delayed eruption of permanent teeth was noticed. Malocclusion with respect to unilateral crossbite on left side and fusion of 81, 82 were observed (Figure 8). The patient's 37 year old father and 15 year old sister also presented with mixed dentitions, with retention of several deciduous teeth, multiple impacted teeth and delayed eruption of permanent teeth (Figures 9a, 9b, 10a, 10b).

On the basis of clinical examination and presence of typical general and intraoral features, provisional diagnosis of cleidocranial dysostosis was made. The patient, his father and sister were advised radiological investigations.

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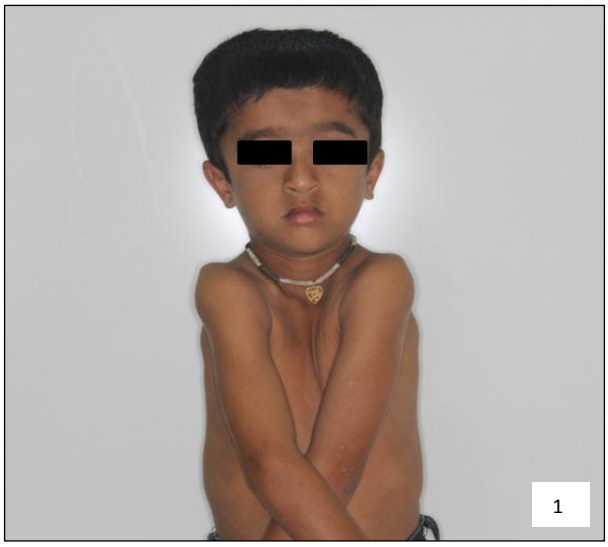
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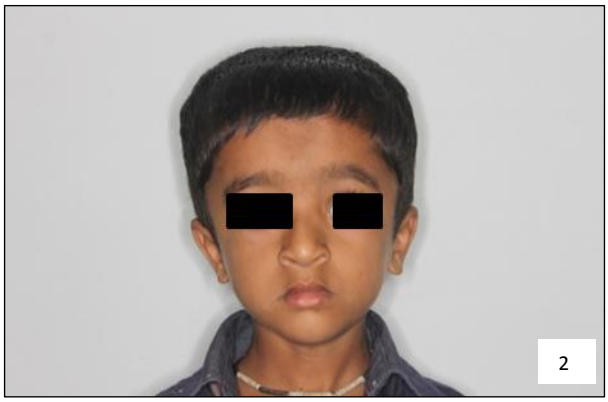
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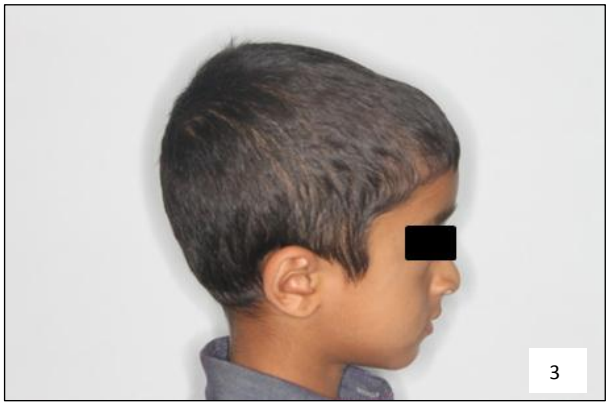
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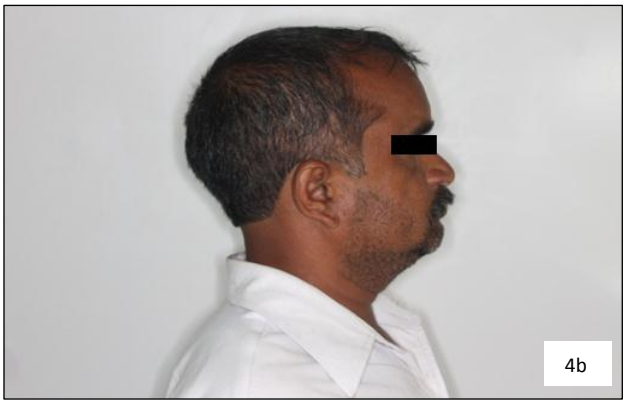
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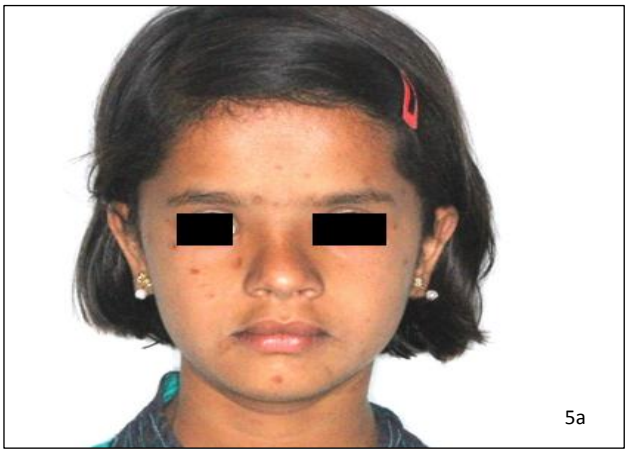
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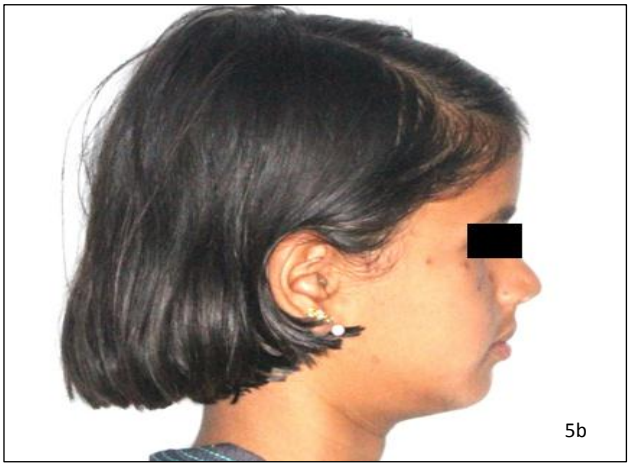
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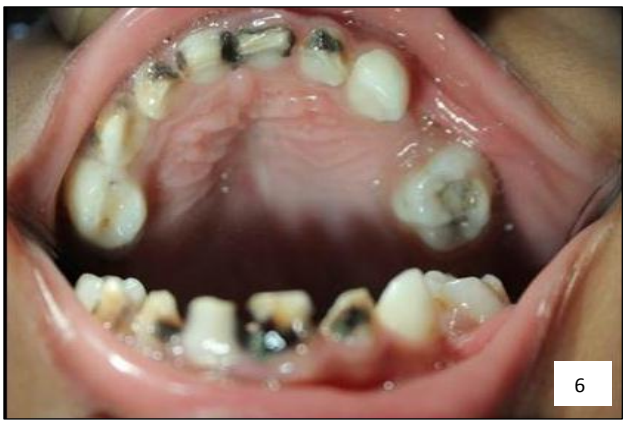
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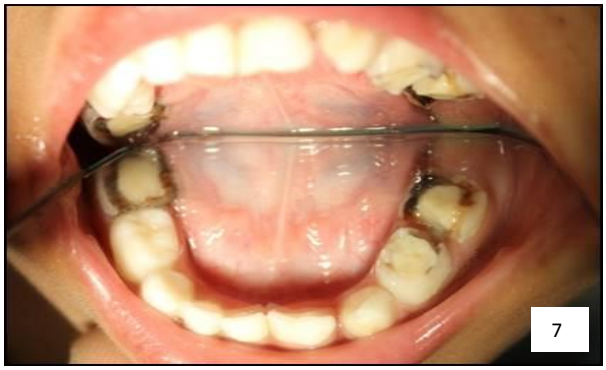
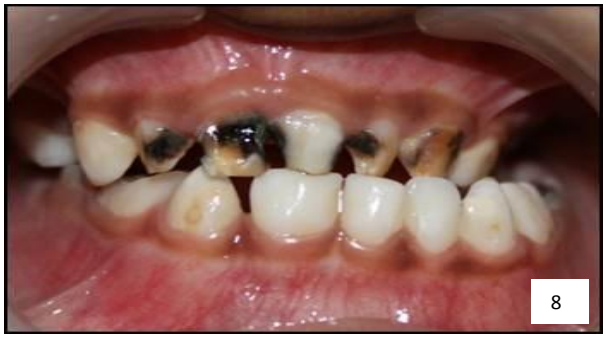
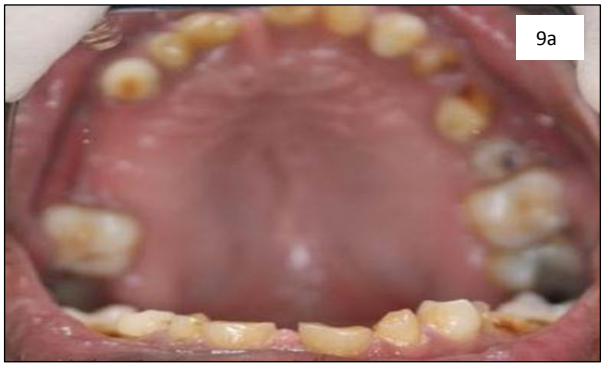
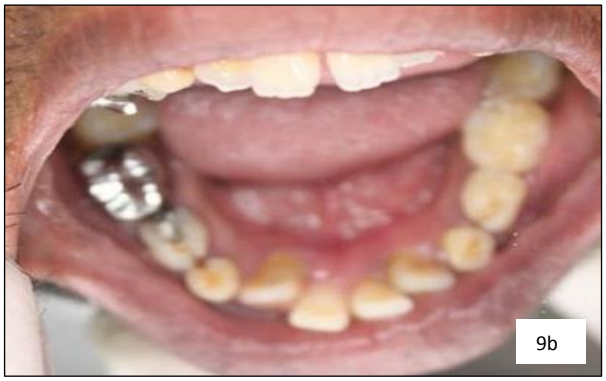
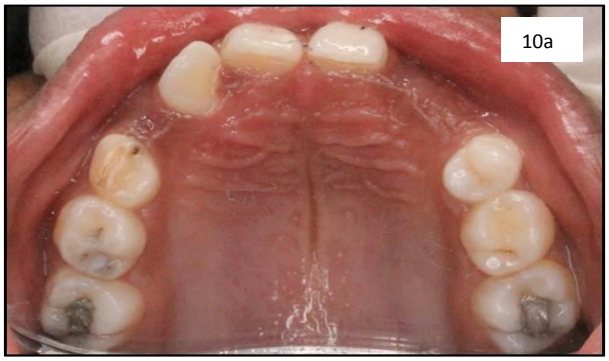
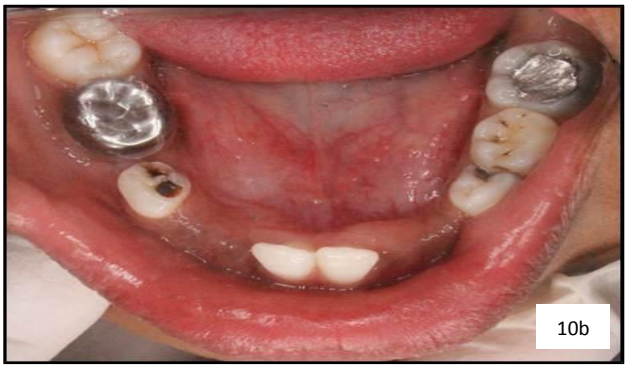


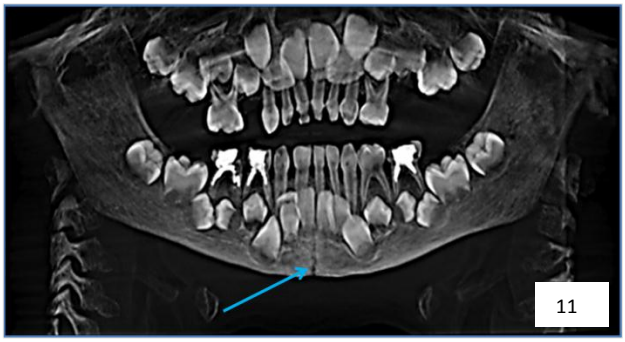


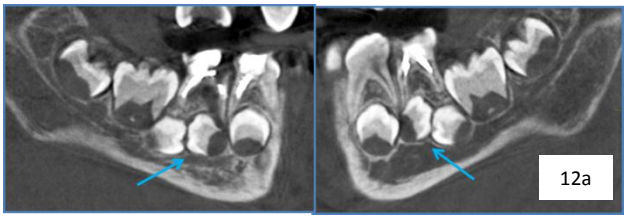








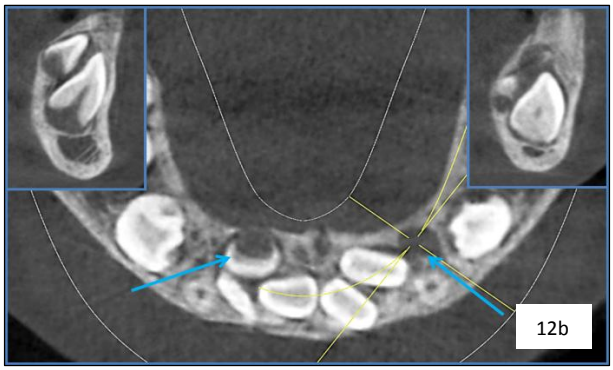




Intraoral periapical radiographs, orthopantomograph, lateral cephalogram, antero-posterior view, chest x-ray and CBCT were advised for the patient, while the father and sister were advised an OPG. The patient's OPG revealed presence of a total of 51 teeth in the jaws, with lack of fusion in the mandibular symphysis (Figures 11). On observing CBCT images, the spatial positioning of the supernumerary teeth in the mandibular bicuspid region bilaterally, lingual to mandibular lateral incisors bilaterally and palatally in relation to 61 was confirmed (Figure 12a, 12b, 12c). The antero-posterior view and lateral cephalogram showed open skull sutures and fontanelles with multiple wormian bones (Figures 13, 14). Chest x-ray revealed a narrow thorax with oblique ribs and hypoplastic clavicles (Figure 15). Also, on panoramic radiographs of the father and sister, retained deciduous teeth and multiple supernumerary teeth were observed (Figures16, 17).

Based on the family history, clinical examination and after the radiographic examination, the diagnosis was confirmed as cleidocranial dysostosis.

A treatment plan was devised for the patient. Accordingly, multiple restorations were done. Pulp therapy was performed on 51, 52, 55, 61, 62, followed up with strip form crowns for the anterior teeth. Stainless steel crowns were placed on 55, 75, 84, 85. Space maintainer was given in the region of 54. The patient is still under follow up care.



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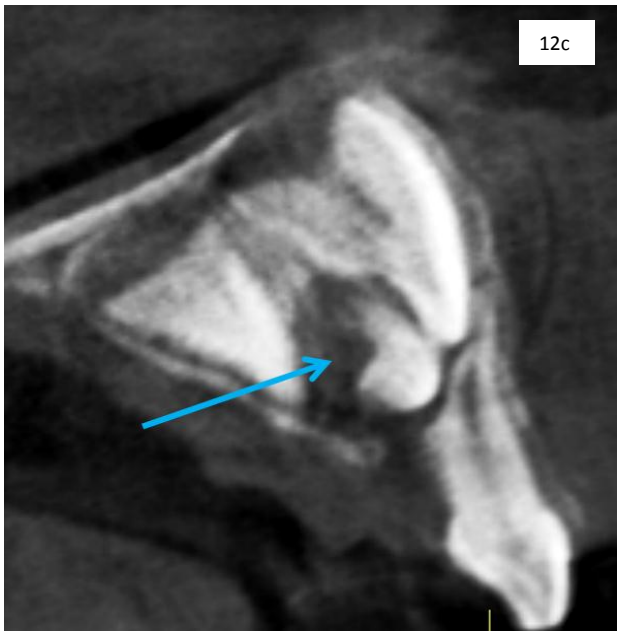

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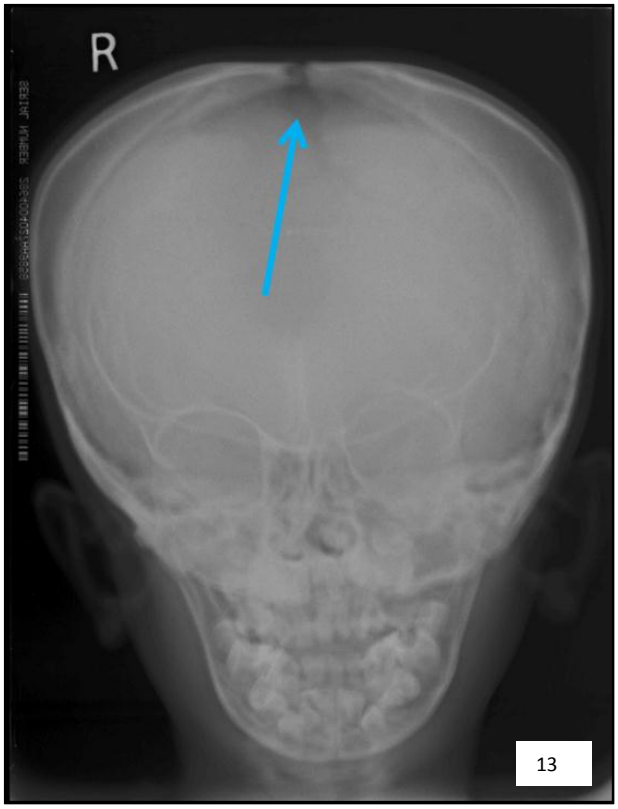

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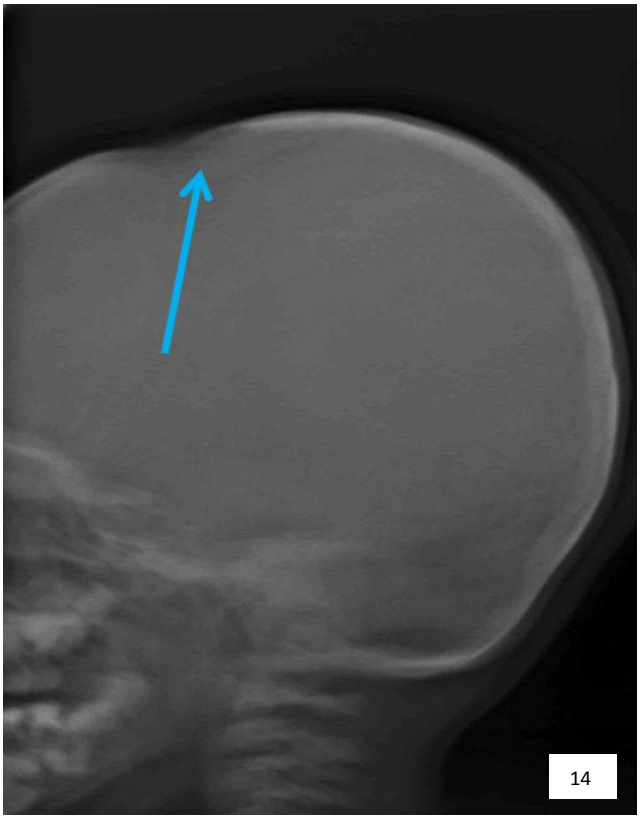

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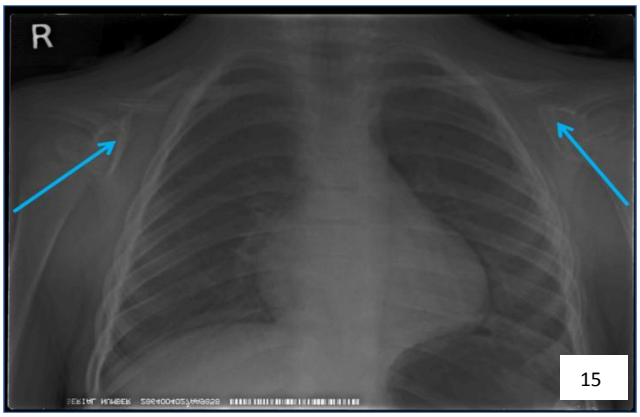

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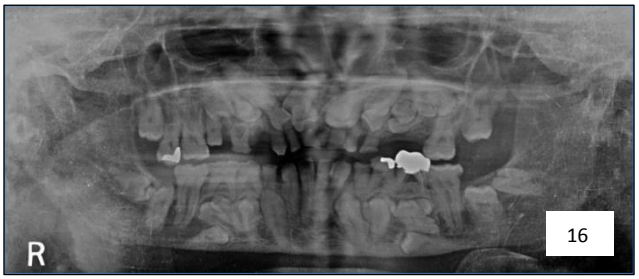

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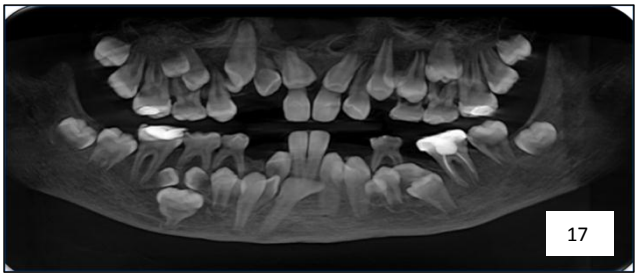

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DISCUSSION

Cleidocranial dysostosis is an autosomal dominant disorder of skeletal tissues having familial hereditary pattern [1,2] as was observed in the present case report. However, all of the three members of the family presented with clinical features with differing degrees of intensity, as is seen with the variable expressivity of cleidocranial dysostosis.[3,4] The prevalence of this condition is 0.5:1,00,000 and incidence, 1:10,00,000. Its earliest history goes back to prehistorical times in Neanderthal skull reported by Greig in 1933. The earliest recognizable report in medical literature has been attributed to Meckel in 1760. The combination of clavicular and cranial defects was recognized by Scheuthauer in 1871. The Parisian physicians Marie and Sainton described its hereditary pattern first and formally named the disorder. In 1908, Hultcranz published a detailed account of anatomical changes. Extensive minor skeletal involvement was emphasized by Jensen and the name of the disorder was changed to 'Cleidocranial Dysplasia'. The RUNX2 gene was mapped to chromosomal locus 6p21 on short arm of chromosome number 6 in 1995.[3] RUNX2 gene plays important role in epithelial-mesenchymal interaction, progressive tooth morphogenesis, histodifferentiation of enamel organ, differentiation of odontoblasts and bone formation. Spontaneous mutation of Cbfa1 molecule on RUNX2 gene causes dysplastic skeletal and dental tissues. There is early developmental disorder of mesenchyme and connective tissue causing delayed ossification of midline structures, particularly membranous bones.

Cleidocranial dysostosis affects males and females equally. The important extraoral clinical features include bossing of calvarial bones, soft area in the region of fontanelles, depression in the midline of forehead, hyperteloric eyes, narrow sloping shoulders, narrow thorax, long neck, short stature and thin built.[5] All of these features were observed in the patient. Hypermobility of shoulders with the ability to oppose them anteriorly, which is a diagnostic feature, was observed in the patient, and his sister. Midfacial hypoplasia, depressed nasal bridge, short terminal phalanges, vertebral abnormalities and pes planus (flat feet) may also be seen, but this was not obvious in this patient. Intraoral features like hyperdontia is common which may lead to dental crowding and malalignment. Malocclusion and narrow high arched palate is also seen. Retention of deciduous teeth and delayed eruption of permanent teeth is a common and significant feature leading to impaction of several teeth. Articulation and mastication may be compromised and the esthetic appearance of the dentition maybe unsightly. This was a cause for concern for the father.

Radiographically, total or partial absence of clavicles is a significant feature in diagnosis. Widened pubic symphysis, vertebral abnormalities, scoliosis, spina bifida, syringomyelia are other abnormalities that can be found. Open skull sutures with large fontanelles and multiple wormian bones are present. The ramus of mandible may appear narrow and parallel with round gonial angles. Abnormalities in coronoid and condylar processes maybe noted. Maldevelopment of sinuses leads to upper respiratory tract infections. Presence of several impacted supernumerary and permanent teeth is a common finding, which sometimes could lead to cyst formation. Lack of eruption of primary and permanent teeth is usually due to decreased eruptive force, and increased odontogenesis and hyperactivity of dental lamina leads to formation of supernumerary teeth. Presence of supernumerary teeth is particularly seen in mandibular premolar and maxillary anterior region.[6] On ground sectioning of teeth, lack of cellular cementum and paucity of acellular cementum is observed, probably causing failure of eruption of teeth.[7] Disorganized dentinal tubules have also been reported in a case. Also, biochemical analysis of these teeth have shown hypophosphatasia with decreased levels of serum alkaline phosphatise.[2]

Several complications can occur in patients with cleidocranial dysostosis, like impaction of permanent teeth, cyst formation related to them, upper respiratory tract infections, otitis media, hearing loss and sinus infections.

CONCLUSION

Along with clinical findings, radiographic evaluation and family history is the most important and reliable means to confirm the diagnosis. Though it is a congenital disorder, findings of cleidocranial dysostosis are often diagnosed later by physicians incidentally while treating for other conditions or usually dentists are first to diagnose the condition. Treatment planning of these patients is affected by various factors. Familial occurrence like in the present case may present with varying degrees of severity, and based on the age of the patient, the treatment needs may vary. However, early diagnosis is of utmost importance to initiate appropriate treatment and achieve desired results successfully. The aim is to establish an esthetic and functional harmony and hence a multidisciplinary approach is required for management on long term basis.

REFRENCES

1. Garg RK and Agarwal P. Clinical spectrum of cleidocranial dysplasia:a case report. BioMed Central. Cases Journal 2008,1:377.doi:10.1186/ 1757-1626-1-377
2. Mohan RPS, Suma GN, Vashishth S and Goel S. Cleidocranial dysplasia: clinico-radiological illustration of a rare case. Journal of Oral Science 2010;52(1):161-166.
3. Roberts T, Stephan L and Beighton P. Cleidocranial dysplasia:a review of the dental, historical, and practical implications with an overview of South African experience. Oral Surg Oral Med Oral Pathal Oral Radiol 2013;115(1):46-55.
4. Porciuncula CGG, de Lira RF, Soares MLL, Araujo DL, Mota LR and Lira LF. Cleidocranial dysostosis: a report on two familial cases. Radiol Bras. 2013 Nov/Dec;46(6):382-384

5. P AR. Cleidocranial dysplasia- A case report and review of literature. NMJ. 2012; 1(1): 35-37
6. Jensen BL , Kreiborg S. Development of the dentition in cleidocranial dysplasia. J Oral Pathol Med 1990;19:89-93.
7. Hemlatha R, Balasubramaniam MR. Cleidocranial dysplasia:A case report. J Indian Soc Pedod Prev Dentistry 2008;26(1)40-43.

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