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Cleidocranial Dysplasia Presenting with Multiple Impacted Teeth: A Case Report and Review of Literature

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Abstract: Cleidocranial dysplasia is a developmental anomaly of the skeleton and the teeth which is result of faulty development of membranous bones. This condition may be inherited or transmitted as dominant characteristics in either sex, or even may appear spontaneously. It presents with a wide range of features, characterized by clavicular aplasia or hypoplasia, short stature, supernumerary teeth. Other features include skeletal abnormalities like skeletal defects of several bones, such as late closure of the fontanels, presence of open skull sutures and multiple wormian bones. The clavicular abnormality ranges from a small defect in one clavicle to complete absence of both. Clinical significance of this condition to dentistry is due to the involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth. Here presenting case of 25-year-old Indian male, presented with the typical features of prolonged retention of deciduous dentition and impacted permanent teeth, mandibular prognathism along with other skeletal abnormalities like shrugged shoulder and the absence of clavicles.
Keywords: Marie and Sainton disease, Supernumerary teeth, Multiple impacted teeth, Open skull sutures.

INTRODUCTION

Among the congenital defects Cleidocranial dysplasia is rare and has autosomal dominance inheritance [1-3]. The defect affects the bone which is undergoing intramembranous assification. Marie and Sainton in 1898[4] gave the first description about this defect. This is also know by other names such as Marie and Sainton Disease, Scheuthauer Marie-Sainton Syndrome and Mutational dysostosis [5] or cleidocranial dysostosis [1]. Spontaneous mutation of gene is associated with this syndrome which encodes for transcription factor core binding factor alpha 1 (Cbfa1) [6]. The gene has been mapped to chromosome 6p21 [7] which is essential for osteoblast and odontoblast differentiation for bone and tooth formation. The related pathology for the condition is due to an early developmental disorder of connective tissue or mesenchyme leading to retarded ossification of bone precursors at junctions. This results in defective ossification or ossification failure in the skeletal structure. Defect in the skeletal structures may be partial or complete absence of the clavicles, presence of open skull sutures, late closure of fontanelles, and multiple wormian bones [2, 8]. Other features include underdeveloped maxilla along with ill-formed paranasal sinuses, and also there is involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth. Because of these clinical features this condition is clinically significant to the dentistry. One more feature of the condition is teeth fail to erupt which is thought to be due to absence of cellular cementum and an increase in the amount of acellular cementum of the roots of the affected teeth [9].

CASE PRESENTATION

A 25-year-old Indian male came to the Department of Oral Medicine and Radiology with the chief complaint of unerupted teeth. History of present illness revealed there were retained deciduous teeth and absence of permanent teeth eruption. Medical history and family history were not relevant. Patient was mentally alert, appeared to be of normal intelligence.

Family history was traced through the patient's parents on back to the maternal grandparents, and cleidocranial dysostosis was not suspected in any of them.

On general physical examination patient was poorly built, short statured, moderately-nourished, with shrugged shoulders with more than normal mobility of the shoulder girdle (Fig. 1). Extra oral examination revealed concave facial profile, frontal bossing, mandibular prognathism, maxillary hypoplasia (Fig. 2) wide nasal bridge, hypertelorism (Fig. 3). On intraoral examination there were only 20 teeth present along with retained 53, 52, 51, 61, 62, 63, 74, 73, 72, 82, 83 and permanent molars present in all the quardrants (Fig. 4). The oral soft tissues appeared normal; the uvula was complete, and there was no macroglossia. The palate was high with maxillary perimeters narrow, large mandibular perimeters.

Based on the history and clinical findings cleidocranial dysplasia was given as provisional diagnosis.

Investigations included panoramic radiograph, postero-anterior projection lateral cephalograph, chest radiograph and genetic mapping.

A panoramic radiograph revealed multiple unerupted permanent teeth and supernumerary teeth in the mandibular and maxillary anterior region (Fig. 5). Posteroanterior view lateral cephalograph revealed wide skull sutures (Fig. 6). The posteroanterior view of a chest radiograph revealed the absence of right clavicle and hypoplastic left clavicle and a bell-shaped ribcage (Fig. 7). Based on these clinical and radiographic findings, a diagnosis of cleidocranial dysplasia was made. However, his chromosomal analysis revealed normal male karyotype 46XY.

A multidisciplinary dental approach involving oral and maxillofacial surgeons, prosthodontist was followed in our case. Written informed consent was obtained from our patient's parents for publication of this case report and any accompanying images.

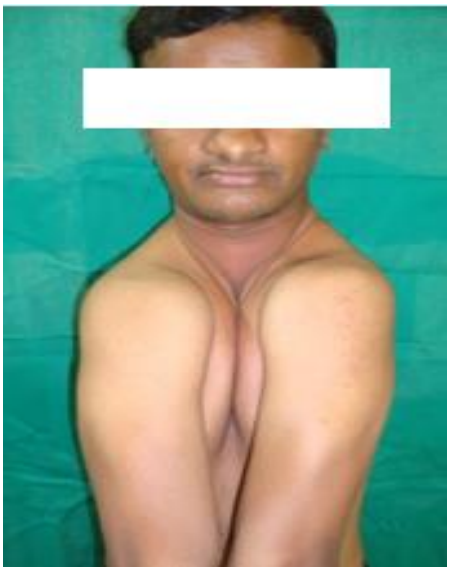

Fig. 1: Photograph showing more than normal mobility of the shoulder girdle

Fig. 2: Extraoral photograph showing concave facial profile, frontal bossing, mandibular prognathism, maxillary hypoplasia

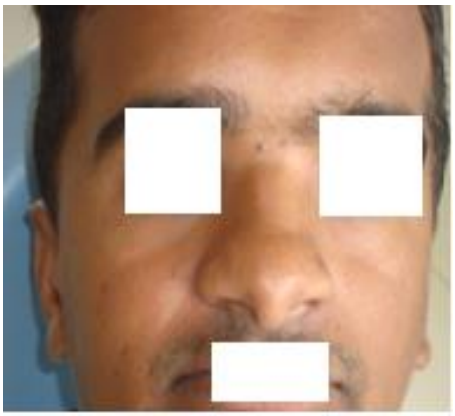

Fig. 3: Extraoral photograph showing wide nasal bridge, hypertelorism

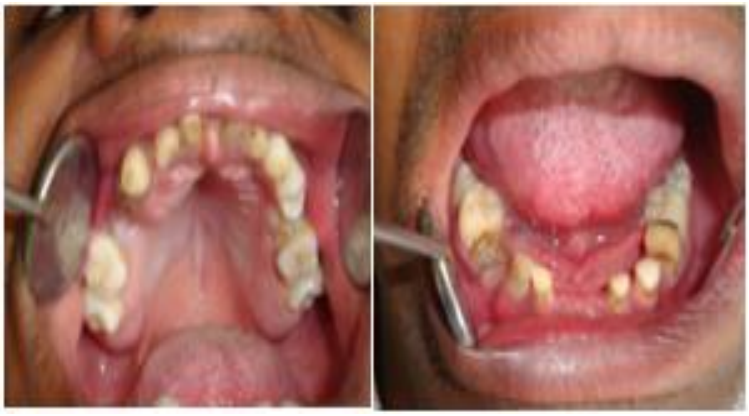

Fig. 4: Introral photograph showing retained deciduous teeth

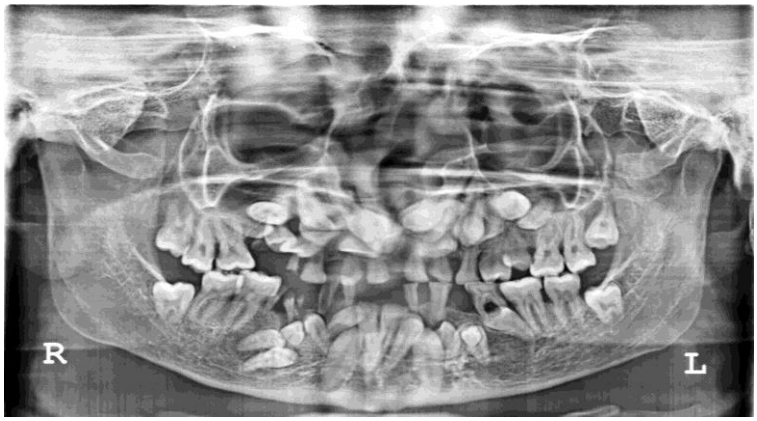

Fig. 5: Panoramic radiograph showing retained deciduous and impacted permanent anterior teeth and supernumerary teeth

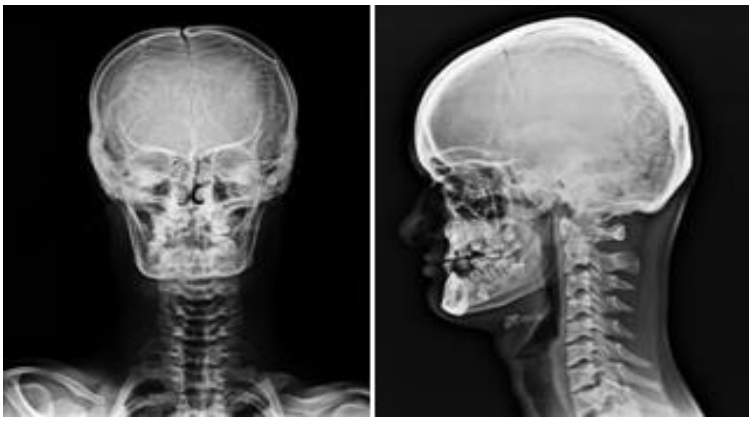

Fig. 6: Posteroanterior view and Lateral cephalogram showing open skull sutures

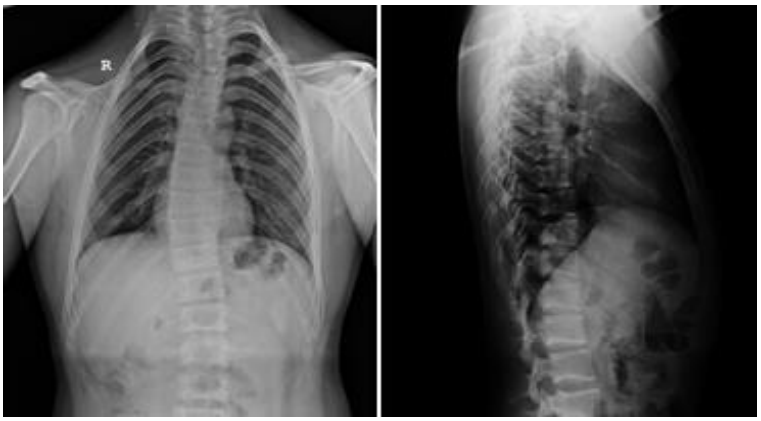

Fig. 7: The posteroanterior view of a chest radiograph revealed the absence of right clavicle and hypoplastic left clavicle and a bell-shaped ribcage

DISCUSSION

Hypermobility of the shoulders, abnormal clavicles, supernumerary teeth and wormian bones are seen to be consistent features of cleidocranial dysplasia. This is a clinical phenotype arising from deregulation of intramembranous and endochondral ossification which may be due to a mutation in Cbfa1. In the present case the features noted were absence of clavicles, broad skull sutures and numerous impacted and supernumerary teeth.

Studies have suggested cleidocranial dysplasia have a point mutation involving *Runx2* in 70% of patients and deletion in 13%. Other test needs to be done in patients whom mutations are not found are the deletion/duplication assay, either Multiplex Ligation-dependent Probe Amplification (MLPA) or, Reverse transcription - quantitative real time polymerase chain reaction (RT-qPCR) [10-12].

It has also been reported that individuals with central core disease could have cytogenetically visible complex chromosome rearrangements [13]. The present case to identify the mutations in the *Runx2*, molecular genetic analysis is recommended as chromosomal analysis and gene mapping were normal.

The characteristic oral findings includes an increase in odontogenesis leading to an excessive number of supernumerary teeth [14], decreased eruptive force of both primary and permanent dentition, prolonged retention of primary teeth [3]. It has been proposed that the supernumerary teeth should be diagnosed and extracted as early as possible so that there will be normal eruption of permanent teeth [15]. Further, an abnormality in the eruption of the anterior teeth interferes with facial aesthetics and lead to other clinical problems.

Both the physical and psychological aspects of the patient were considered in the treatment planning. Prosthetic rehabilitation was done by giving the crowns for individual teeth. Simultaneously, the psychological counseling and behavior management methods were taken into consideration ultimately resulting in an improvement in the self-image and confidence.

CONCLUSION

Patients with Cleidocranial dysplasia have compromised aesthetics usually seen as an unexpected event in the course of observing or treating a patient. Early diagnosis helps in 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 the psychological aspects. As a dentist it is our duty to correct the aesthetics of the patient which enhances confidence and self esteem of the patient.

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