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ORAL MANIFESTATIONS OF ACHONDROPLASIA: A CASE REPORT

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ABSTRACT

Achondroplasia a common form of dwarfism, caused by a single recurrent point mutation in more than 97% of patients, is an autosomal dominant disorder with an incidence of approximately 1/7500. The name of this disease was called Chondrodystrophia foetalis before Parrot in 1878 reported the name of this disease as Achondroplasia, distinguished from other similar diseases. The present case report deals with a patient who was diagnosed with achondroplasia. The craniofacial features are discussed and the management done and planned for further management has been discussed.

KEY WORDS:. **Achondroplasia, short limbed dwarfism, malocclusion, Mid face hypoplasia**

INTRODUCTION

The term achondroplasia was first used by Parrot in 1878. The incidence of this condition varies from one in 26 000 to one in 66 000 births, but it is one of the most common types of dwarfism.[1] Apart from small stature and short limbs, the affected children had good general health, normal intelligence and buoyant personalities.[2] It is characterized by disproportionate shortening of the extremities especially the proximal segments, a large head with a prominent forehead, and midfacial hypoplasia.[3] The dental findings include undergrowth of the base of the cranium with visible cephalometric finds, anterior open bite, slightly larger tongue and the size, number, form and growth of the teeth were usual.[4] Craniofacial malformations include an enlarged calvarium with hydrocephaly and frontal bossing, a depressed nasal bridge, a short posterior cranial base, a small foramen magnum, a retrognathic maxilla and a normal mandible. [5] The major abnormality found in the growth plate of achondroplasia and hypochondroplasia is quantitative rather than qualitative. Indeed, the molecular defect in these two skeletal dysplasias now has been identified as a point mutation in the fibroblast growth factor 3 receptor gene. [3, 6]

Although the clinical features and natural history of achondroplasia have been documented extensively, there is a paucity of information on the dental manifestations. For instance, in a multicentre review of 193 affected persons, only brief mention was made of potential dental problems. [7] There are few studies in the literature reporting the dentofacial findings in achondroplasia, even though it is not a rare condition and its molecular basis has recently been discovered. [1,4,8]

Case report

A 9 year old male patient was referred to the Department of Pedodontics with the complaint of decayed and irregularly placed teeth. He had been diagnosed by achondroplasia immediately after birth. At around 6 years of age he had undergone bilateral lengthening of the limbs. The patient appeared to be well built, healthy and intelligent. He was 87cms tall at the initial visit **[Fig.1].** Typical of achondroplasia he had a concave profile, large calvarium with a bulging forehead and saddle shaped nose **[Fig.2].**

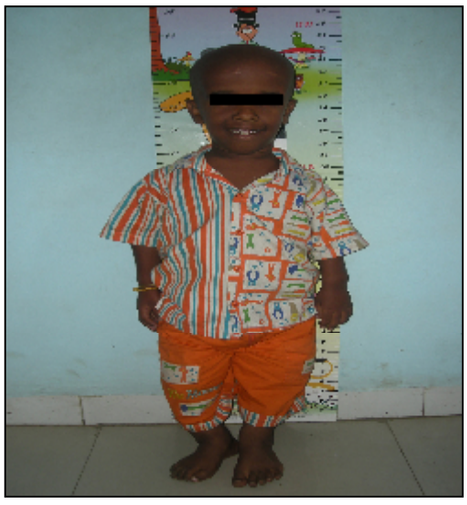

Fig.1. short stature of the patient

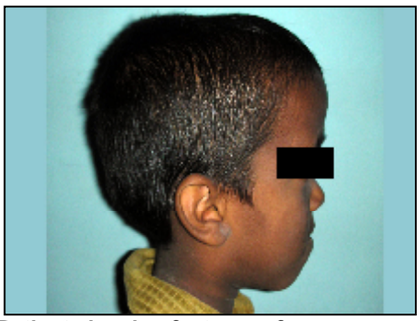

Fig.2 Patient showing features of concave profile, large calvarium with a bulging forehead and saddle shaped nose

Medical examination included radiographic investigation of the head including lateral view **[Fig.3]** confirming large calvarium, frontal bossing and PA view **[Fig.4]** revealing midface hyoplasia. Radiographs of the thoraco-lumbar region showed the characteristic features of patients with achondroplasia. Intraoral findings reveal unusual large tongue, crowding of mandibular anteriors and posterior cross bite **[Fig.5]** and a class III skeletal malocclusion that was also revealed in the cefalometrics **[Fig.6].** The Panoramic view revealed no abnormalities in the size and shape of the teeth [**Fig.7].** Multiple carious lesions were observed **[Fig.8 and Fig. 9].** Primary maxillary central incisors were retained thus leading to labial eruption of permanent successors. The patient had undergone extractions of right and left upper deciduous first molar two years earlier. Dental management involved extraction of retained deciduous dentition and restoration of carious teeth with GIC FUJI IX **[Fig.10 and Fig. 11].**

Discussion

In more than 97% of patients with ACH, the disease is caused by a single, recurrent point mutation resulting from a G to A substitution at nucleotide position 1138 G1138A in exon 10 of the fibroblast growth factor receptor 3 gene FGFR 3. Moreover, in more than 90% of cases the condition is sporadic and represents a spontaneous germline mutation. [9, 10]

In Achondroplasia, dwarfing is presumably related to slow enchondral bone formation since intra-membranous and periosteal ossification is normal. The characteristically recessed face and nose of the patient with Achondroplasia have been explained as resulting from the stunted growth of the bones of the base of the skull and face , bones which from in part from cartilage, in contrast to the calvarium which ossifies in membrane and hence develops normally. The discrepancy between the short extremities and the comparatively long trunk of the achondroplastic patient and his peculiarly shaped pelvis has not been satisfactorily explained. [11, 12] The affected children had mouth breathing because of upper-airway obstruction. The teeth are in proper shape, with normal

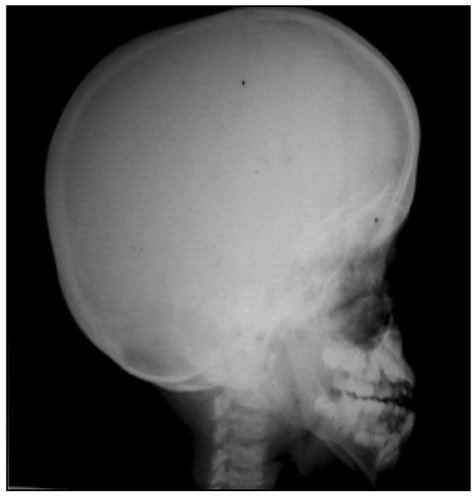

Fig.3 Radiograph of head [Lateral View] revealing large calvarium

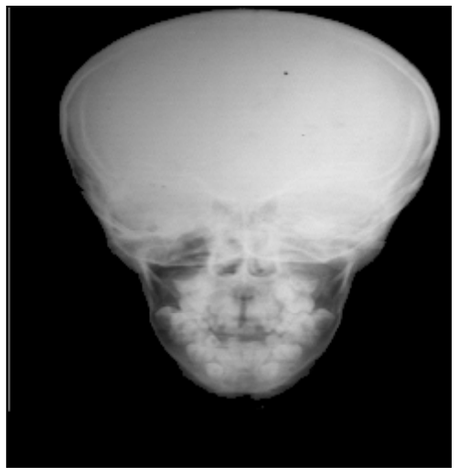

Fig.4 Radiograph of head [PA View] revealing midface hyoplasia

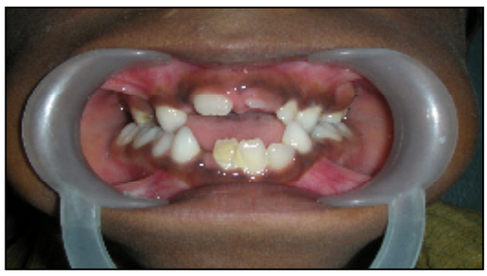

Fig.5 Irregularly place teeth, crowding of mandibular anteriors

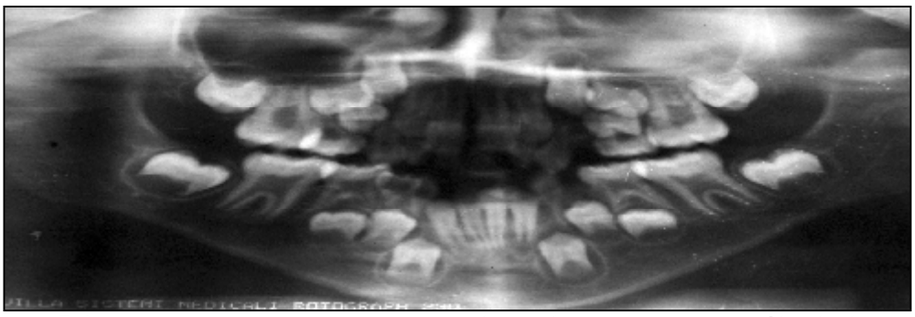

Fig.7 Panoramic view revealed no abnormalities in the size and shape of the teeth

texture and mechanical properties with or without malalignment. The mouth is wide, and the upper dentition prominent. Wide spacing of the teeth, which is often prominent in adulthood, was not seen in the children. Short limbs, stunted stature and occasional spinal malalignment made it difficult for the children to sit comfortably in a conventional dental chair. Chronic backache may compound this problem. Upper airway obstruction due to depression of the nasal bridge, variable choncal atresia and enlarged adenoids results in mouth breathing. A small nasal pharynx and larynx posed additional problems for anesthesia. The possible presence of cranio-cervical instability necessitated special precautions in head control during dental intervention. A large heavy head, sometimes with arrested hydrocephalus and an implanted shunt, made head control difficult. [2]

According to Dunbar *et al*. the main orthodontic problem in achondroplasia is class III malocclusion consequent upon inherent shortening of the base of the skull; this abnormal configuration leads to retraction and decrease in vertical height of the maxilla.[13] Many skeletal malocclusions can be treated by using pre-pubertal and pubertal growth phases. In achondroplastic patients, however, the treatment choices may be limited since growth potential cannot be used in the same way. The presence of macroglossia also limits the treatment modalities by contraindicating tooth extraction. The management of the patient as planned by our department includes orthodontic correction with expansion appliance followed by head gear therapy. We will not be considering taking advantage of the growth phase period but will consider Short term Recombinant Human Growth Hormone Treatment. Our treatment plan will include a 1 year human Growth hormone Therapy LG Eutropin Inj as per the treatment duration in the study done by Shohat M et al who demonstrated a greater growth acceleration of the severely affected areas i.e. the upper and lower limbs. [14]

Fig.8 Multiple carious lesions in maxillary segment

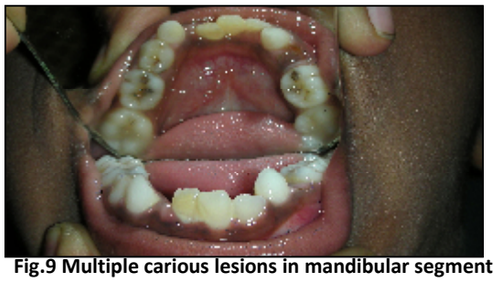

Fig.9 Multiple carious lesions in mandibular segment

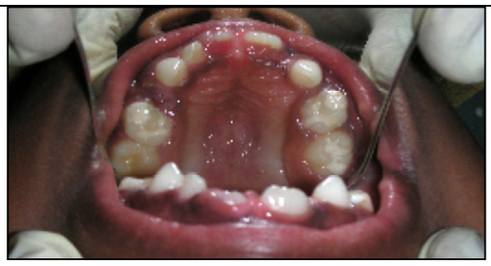

Fig.10 Oral prophylaxis, extractions and restorations with Fuji IX done -maxillary segment

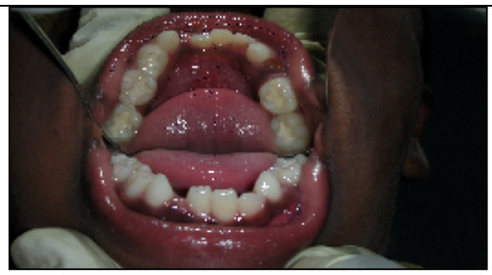

Fig.11 Oral prophylaxis, extractions and restorations with Fuji IX done- mandibular segment

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