CASE REPORT

Achondroplasia: A Rare Syndrome

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Abstract

Achondroplasia also known as "chondrodystrophia fetalis" is a rare disorder that affects the growth of long bones and cartilages. This disorder is characterized by dwarfism with marked disproportionate short limbs. Considering the complications that may emerge during the treatment of a patient with achondroplasia, it is pertinent that the pediatric dentist should be familiar with the marked features of this syndrome as dental management is duly restricted by practical problems associated with this condition. Patients with achondroplasia not only require specific medical management but also need special attention with definite dental management along with psychological support to help them lead a normal life and cope up with the medical and social challenges of life. We report in this paper about children aged between 3 years and 9 years with achondroplasia with extremely short stature, frontal bossing, macrocephaly, and various dental problems.

Keywords: Achondroplasia, Concave facial profile, FGFR-3, Trident hands.

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Introduction

Achondroplasia is a non-threatening type of autosomal dominant condition. It is the most common type of short-limb dwarfism, occurring 1 in 25,000 births.[1] Prevalence rate is around 1:10,000 to 1:50,000.[2] Most cases exhibit gene mutilation for fibroblast growth factor receptor-3 (FGFR-3).[3] This leads to a drop in calcification of endochondral tissue, constrained growth of cartilage cells, reduced cellular hypertrophy, and decreased cartilage matrix formation. An unbalanced growth pattern between endochondral bone and the underlying organs sets off several orthopedic, neurological, respiratory, and dental issues in these children. Recurrent ear infections, cor pulmonale, and dyspnea are frequent concerns. It is life-threatening, usually during the initial years of life, because of respiratory insufficiency caused by severe deformity of the rib cage. Prenatal diagnosis of achondroplasia with routine ultrasound shows the images of shortened long bones. Besides, molecular testing (FGFR-3 mutational testing) of prenatal specimens can be done as a confirmatory test.[4] The diagnosis can usually be made based on clinical characteristics and specific features on radiographs.

Case Description

This is a case series of three children from both genders aged between 5 years and 10 years with a known medical history of achondroplasia reported to our department for a routine dental check-up. Familial history revealed their parents to be related as first degree cousins. The patients developed hydrocephaly at birth. They also exhibited delayed motor milestones due to hypotonia and mechanical difficulty in balancing the large head and short extremities. These patients started walking after 2 years of age. They also had a recurrent ear infection, tonsillitis, and otherwise appeared to be well adjusted and intelligent. Extraoral examination revealed classical manifestations associated with achondroplasia such as short stature, short stubby trident hands (Fig. 1), frontal bossing, saddle nose, and midfacial hypoplasia (Fig. 2). A concave facial profile was observed (Fig. 3).

Intraoral examination in these patients revealed mixed dentition. The size, number, and form of teeth were normal with multiple carious lesions (Fig. 4). Other features seen were

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Fig. 1: Trident hands

Fig. 2: Achondroplastic patient with short stature with shortening of the arms and legs

Fig. 3: Concave facial profile

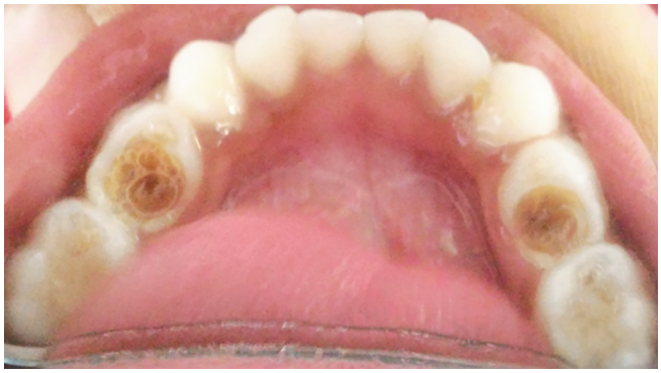

Fig. 4: Multiple carious lesions

macroglossia, tongue-thrust swallowing, generalized gingivitis, anterior reverse jet, anterior open bite, delayed dental development due to altered bone growth and crowding. The maxillary incisors were proclined and the mandibular incisors were normally inclined with Angle's Class III molar relation. Radiographic findings showed prognathic mandible and compressed cervical vertebrae (Fig. 5). Dental treatment performed included oral prophylaxis, topical fluoride application, pit and fissure sealants, and glass ionomer restorations, pulpectomy followed by stainless steel crown based on individual treatment needs. The patients were cooperative and advised to come every 6 months of follow-up.

Discussion

Achondroplasia is a benign form of chondrodysplasia which means "without cartilage formation". Most cases result from a spontaneous mutation of a single gene *fgfr*-3 on chromosome 4. Fibroblast growth factor receptor-3 also plays an important role in cell growth and division, determination of cell type, the formation of blood vessels, wound healing, and embryo development. The mutation causes the receptor to become activated in the absence of a physiologic ligand which accentuates normal receptor function of negatively regulating bone growth.[5]

Individuals with achondroplasia progressively fall below normal standards for length and height. The average height for women is 112–136 cm and for men is 118–145 cm. The limbs are relatively shorter than the trunk with short upper arm and thighs. The head is generally large with a prominent forehead and flat nasal bridge. Fingers and toes are short with trident hands and feet that appear small and wide (Figs 1 and 2). Special growth curves for chest circumference of patients with achondroplasia from birth to 7 years are available. Three distinct phenotypes exist: phenotypic group I patients possess relative adenotonsillar hypertrophy, group II have muscular upper airway obstruction and progressive hydrocephalus, and group III patients have upper airway obstruction and without progressive hydrocephalus. Kyphoscoliosis may develop during infancy[5] (Table 1).

Table 1: Clinical features and orofacial manifestations of patients with achondroplasia[6]

Clinical features	*Orofacial manifestations*
Disproportionately short stature. Girls grow to a height of about 126 cm (4 feet 2 inch) and boys to a height of 131 (4 feet 4 inch).	Macrocephaly, frontal bossing, midfacial hypoplasia, relative mandibular prognathism, flat nasal bridge.
Rhizomelic shortening of the arms and legs with trident configuration. Polydactyly may be present.	Retarded eruption of permanent teeth.
Thoracolumbar protuberance, lumbar lordosis, limitations at the elbow joint.	Crowding, narrow maxilla leading to skeletal and dental Class III malocclusion and crossbite.
Short and round iliac bones, narrow greater sciatic notch, and decreased interpedicular distance from L1 to L5 are seen.	
Normal intelligence.	

A rare complication is a cervicomedullary compression (compression of the upper spinal cord at the base of the brain) which can present with episodes of apnea which can be confirmed by a CT or magnetic resonance imaging (MRI) scan and surgical intervention. In the adolescent and adult, compression of nerve roots in the spinal canal may occur with the sensation of numbness or weakness in the legs. Although shunt was reported to be placed in some achondroplastic children due to hydrocephalus which makes head control difficult and may necessitate antibiotic prophylaxis before dental treatment.[7–9]

The various differential diagnoses for achondroplasia with respective clinical features have been described in Table 2.

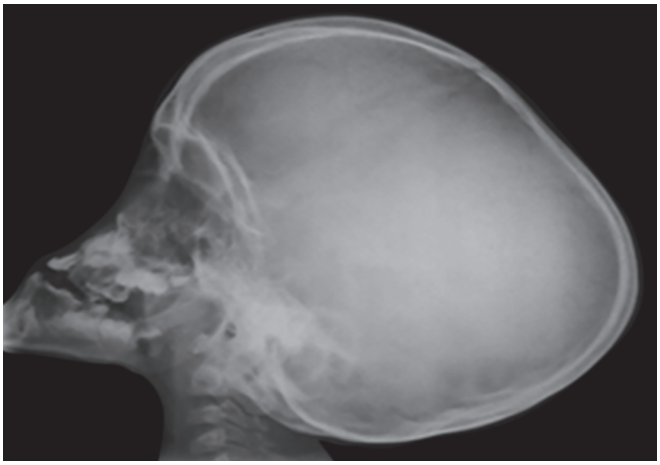

Fig. 5: Prognathic mandible along with constricted foramen magnum and compressed cervical vertebrae

Table 2: Differential diagnosis of achondroplasia with clinical features

Differential diagnosis	*Clinical features*
Hypochondroplasia	Short stature; stocky build; disproportionately short arms and legs; broad, short hands and feet; mild joint laxity, shortened ilia.[10]
Turner's syndrome	Short stature, short neck, a broad chest, genu valgum, and nail dysplasia.[11]
Leri–Weill syndrome	Madelung wrist deformity, mesomelia, and short stature.[12]

Dental management of achondroplastic children requires special considerations because of the characteristic feature of disproportionate and short stature which can lead to a wide range of psychological and social problems. In addition to short limbs, stunted growth and occasional spinal mal-alignment make it difficult for achondroplastic children to sit comfortably on a conventional dental chair. Thus, the dental chair was lowered, and a step stool was used to help the patient to get on the dental chair easily. Furthermore, special precautions in head control like a neck pillow, backrest, and cuddle jackets during the dental intervention (Fig. 6) are essential due to the possible presence of craniocervical instability, foramen magnum stenosis, and limited neck extension which may result in spinal injury.

General anesthesia (GA) considerations pose certain complications due to anteriorly placed epiglottis, small nasal pharynx and larynx, difficulty in intubation, lumbar lordosis, narrowing of the spinal cord, and small chest.[13,14] Dental professionals need to recognize various risk factors and potential complications before administering sedation or anesthesia. When dental treatment is planned under GA, it is recommended to perform a radiologic evaluation of foramen magnum, airway evaluation, preoxygenation before anesthesia, use of appropriate endotracheal tube size, oral intubation, and administration of oxygen after extubation. If cervicomedullary compression is not suspected, one must determine how far the patient can extend his or her neck. In anticipation of difficult intubation, alternative techniques (such as blind nasal, flexible fiberoptic, or retrograde intubations) should be considered.

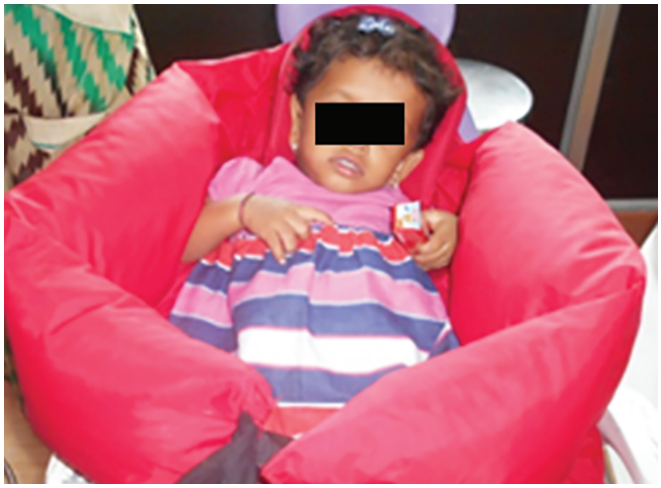

Fig. 6: Special intervention for dental procedure

Table 3: Dental management protocol[6]

1. Detailed case history should be taken.
2. Special psychological behavior management is mandatory.
3. Conductive hearing loss may be present. The hearing aid should be adjusted to prevent amplification of sounds in the dental operatory.
4. Delayed eruption of permanent teeth may be seen. An investigation is warranted to determine whether the teeth are missing or eruption is delayed. All children with this condition should be evaluated for orthodontic needs by 5 or 6 years as Class III malocclusion is of concern.
5. Shunt placed due to hydrocephalous causes difficulty in head control during dental treatment. Precaution should be taken before dental treatment to stabilize the head as there is a possibility of foramen magnum stenosis, limitation in neck extension which can lead to respiratory complication. Antibiotic prophylaxis before treatment becomes necessary.
6. It is advised to perform dental treatment under local anesthesia, because general anesthesia poses certain complications due to anteriorly placed epiglottis, small nasal pharynx and larynx, difficulty in intubation, lumbar lordosis, narrowing of the spinal cord, and a small chest.
7. Based on the treatment needs of the patient, dental procedures can be carried out.

Thus, a patient with achondroplasia not only requires specific medical management but also special attention with customized dental management along with psychological support to help them lead a normal life and cope up with the medical, dental, as well as social challenges of life. There are few studies in the literature reporting the dentofacial findings in achondroplasia, as it is a rare condition. These case series describe the craniofacial, dentofacial findings and potential dental problems and various approaches taken into consideration while delivering the preventive treatment measures like diet counseling, oral prophylaxis, topical fluoride application, pit and fissure sealants with utmost precautions that are necessary in the management of achondroplastic patient. A summary of protocols to be followed has been mentioned in Table 3.

Conclusion

This case series describes the characteristic features of achondroplasia that may lead to respiratory, neurological, skeletal, dental, and psychosocial problems. A pediatric dentist should be able to identify the condition, understand clinical features, emphasize maintaining oral health by instituting preventive measures like diet counseling, topical fluoride applications, pit and fissure sealants, vigorous oral hygiene measures, monitor craniofacial growth and development, correct jaw relationships and dental occlusion for proper function and improved esthetics, thereby instilling a positive dental attitude.

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