Title “Craniofacial Manifestations Of Achondroplasia ”


ABSTRACT

Achondroplasia is an autosomal dominant disorder and it is the most common form of dwarfism with short limb. Achondroplasia is caused by mutation in the fibroblast growth factor receptor-3 gene (FGFR3). Craniofacial features include enlarged calvarium with hydrocephaly, brachycephaly, midfacial hypoplasia, depressed nasal bridge, prominent forehead dental malocclusions and delayed tooth eruption. Less often, infants and children may have serious health consequences related to hydrocephalus, craniocervical junction compression, upper-airway obstruction, or thoracolumbar kyphosis. Anticipatory care should be directed at identifying children who are at high risk and timely intervention is needed to prevent serious sequelae and thus multidisciplinary approach is highly essential.

Keywords: Achondroplasia , Craniofacial manifestations , dwarfism , mutation.
INTRODUCTION:

Achondroplasia, is an autosomal dominant hereditary skeletal disorder caused due to failure of normal conversion of cartilage into bone that begins during fetal life. It is also known as chondrodystrophiafetalis (Gorlin et al 1990). Achondroplasia is a nonfatal form of chondrodysplasia.\(^1,2\) It affects about one in 20,000 infants or one in every 40,000 children globally. Approximately 150,000 persons have achondroplasia worldwide.\(^3\) There are more than 10,000 individuals living with achondroplasia in the United States (equal frequency in men and women and across all racial and ethnic groups) making it the most common skeletal dysplasia.\(^3\) Achondroplasia is caused by mutation in the gene that codes for the fibroblast growth factor receptor type 3 (FGFR3) which is located at 4p16.3.\(^4-6\) The affected individuals may have disproportionate extremities, craniofacial features include prominent forehead, midfacial hypoplasia, foramen magnum stenosis, low nasal bridge, and dental malocclusion with anterior open bite. Apart from this physical deformity, the individual bears a normal intelligence level (Stephen et al 2005).\(^7\) The purpose of this case report is to present the craniofacial features of achondroplasia and discuss special consideration that should be taken in the case of dental management of this condition.

CASE REPORT:

A 7 years old female patient accompanied by her mother reported with the chief complaint of stains in the lower right and left back teeth since 6 months. Patient was born to consanguineous parents. Her mother, father and maternal grandmother are short statured.

The natal and post-natal history revealed that the patient was delivered through vaginal delivery at 8.5 months of gestation. Post natal physiological events were in normal range. The patient appeared to be well adjusted, healthy and intelligent. The motor developmental milestones, social and adaptive milestones and language milestones were reportedly normal according to her mother. Patient’s mother was counseled to get aborted by her gynecologist at her third month of gestation.
Anthropometry revealed a weight and height of 11.8 Kg and 88 cm respectively. Mesomelic dwarfism with upper limb to lower limb ratio of >1 (expected ≤1). Patient appeared to be well-adjusted, other general examination findings included frontal bossing of skull, hyperteloristic eyes, leptoprosopic face, saddle nose and rhizomelic shortening of the arms and legs. A concave facial profile was also noticed. Her vital signs were within normal range.

Intraoral examination revealed mixed dentition. Dental caries was present in 54, 64, 74, 75, 84, 85. The size, number and form of teeth were normal with Class I Molar relationship on both sides. The periodontal status appeared to be normal. The lateral cephalogram revealed reduced length of the base of the skull, brachycephaly and mid facial hypoplasia. The panoramic radiograph revealed radiolucent areas in the crown areas of 54, 64, 74, 75, 84, 85. A hand wrist radiograph was also taken which showed nearly equal length of metacarpals which was ‘STARFISH’ in shape. The diagnosis was
finally confirmed as Achondroplasia by considering all the clinical and radiographic features. Treatment plan was formulated to restore all the carious teeth and the oral prophylaxis was done for the patient as the oral hygiene of the patient was found to be satisfactory. The patient is being motivated for medical intervention and has been kept on regular follow up.

**DISCUSSION**

Achondroplasia is a non-lethal form of chondrodysplasia. It means “without cartilage formation” (Chawla et al 2012). In comparison with achondroplasia the changes and severity are milder in hypochondroplasia and more severe and lethal in thanatophoric dysplasia (Trotter and Hall 2005). Dental development are mostly delayed in achondroplastic children because of altered bone growth (Vaccaro and Albert 2001). Dealing with achondroplastic children needs special attention with special psychological management during dental treatment, as the presence of disproportionate short stature can cause a number of psychosocial and social problems (Trotter and Hall 2005). Presence of short stature and short limbs, in addition to chronic backache will make achondroplastic patient to sit uncomfortably on a conventional dental chair (Hunter et al 1998). Backache usually indicates spinal stenosis which is noticeable at late childhood and early adolescence of achondroplastic patient. Lowering the dental chair and using step stool will help the achondroplastic child to get on the dental chair easily (Hecht and Butler 1990). A cushion behind the child’s back may be required during dental treatment for good posture and to reduce back pain, it will also enhance the comfort level of the patient. It is advised to perform the dental treatments under local anesthesia. General anesthesia may pose certain complications due to small nasal pharynx and larynx, anteriorly placed epiglottis, difficulty in intubation, lumbar lordosis, narrowing of spinal cord and small chest. (Kalla et al 1986). Hyperextension of neck should be avoided to prevent cord compression because foramen magnum stenosis is common. Delay in the acquisition of speech is a recognized complication in achondroplasia. The American Academy of Pediatrics (AAP) guidelines recommend speech evaluation by 2 years of age and it is part of their anticipatory care. Requirement of drugs including inducing agents and relaxants is judged on body weight basis (Morrow and...
black 1998)\(^{17}\). It is important for dental professionals, including oral surgeons, pediatric dentists and orthodontists treating these patients, to recognize risk factors and potential complications before sedation or anesthesia. It is also highly recommended to do radiologic evaluation of foramen magnum, preoxygenation before anesthesia, using appropriate endotracheal tube size, oral intubation and administration of oxygen after extubation. (Barone and Eisig 1994)\(^{18}\). Mouth breathing due to upper-airway obstruction is a constant feature in achondroplastic children (Onodera et al 2005)\(^{19}\). Although there is no cure for achondroplasia, extended limb lengthening has been used to improve stature (Aldegheri and Dall’Oca 2001)\(^{20}\).

**CONCLUSION**

On comparing the clinical and radiographic features presented by this patient with the literature available on achondroplasia, we found all the findings similar. Hence, a patient with achondroplasia not only requires medical management but also requires specific dental management along with psychological support to help them lead a normal life and cope up with the medical as well as social challenges of life. Dentists treating these children should be able to recognize these features and its complications. Patient can also be encouraged to join special support group therapy for more social interaction.

**REFERENCES:**


Correspondence: Juhi Jahan, A
Postgraduate student, Department of Oral Medicine and Radiology,
Indira Gandhi Institute Of dental Sciences,
MGMCRI Campus, Pilliyarkuppam,
Pondicherry- 607402.
Email: juhi.omr@gmail.com; contact no: 8015489566

Co-author (1): Vandana S
Readers, Department of Oral Medicine and Radiology,
Indira Gandhi Institute Of dental Sciences,
MGMCRI Campus, Pilliyarkuppam, Pondicherry-607402.
Email: drvandanapavit@gmail.com; contact no: 9442368880

Co-author (2): Vishwanath Rangdhol
Professor and Head, Department of Oral Medicine and Radiology,
Indira Gandhi Institute Of dental Sciences,
MGMCRI Campus, Pilliyarkuppam, Pondicherry-607402.
Email: vishident@gmail.com; contact no: 9448076396

Co-author (3): Santosh Palla
Postgraduate student, Department of Oral Medicine and Radiology,
Indira Gandhi Institute Of dental Sciences,
MGMCRI Campus, Pilliyarkuppam, Pondicherry-607402.

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