Short communication

iMedPub Journals
www.imedpub.com

Global Journal of Research and Review
ISSN 0975-9344

2021
Vol.8 No.2:66

Achondroplasia in a 8 Year Old Male Child: Report of a Rare Case

Priyanka Mayer*

Department of Oral Medicine and Radiology, Chhatrapati Shahu Maharaj Shikshan Sanstha Dental College and Hospital, Aurangabad, Maharashtra, India.

***Corresponding author:** Priyanka Mayer, Department of Oral Medicine and Radiology, Chhatrapati Shahu Maharaj Shikshan Sanstha Dental College and hospital, Paithan Road, Kanchanwadi, Aurangabad-431 002, Maharashtra, India. Tel: 07033013321; E-mail: khamboo1927@gmail.com

Received date: January 13, 2021**; Accepted date:** January 26, 2021**; Published date:** February 3, 2021

Citation: Mayer P (2021) Achondroplasia in a 8 Year Old Male Child: Report of a Rare Case. Glob J Res Rev Vol. 8 No.2:66.

Abstract

Achondroplasia is considered as the commonest cause of dwarfism, which is inherited as an autosomal dominant trait. It has a prevalence of 1 in 15,000 to 40,000 live births. This genetic disorder usually manifests at birth as a disproportionately enlarged head in comparison to the body, and a reduced size of arms and legs. Here, we report a case of an eight year male patient, with features of achondroplasia, who reported to us with a lack of eruption of upper front tooth since past 1 year.

Keywords: Achondroplasia;Dwarfism

Case Report

An eight-year-old Indian male child, reported to the Department of Oral Medicine and Radiology, with a complaint of missing upper front tooth since past 1 year. Patient's father revealed, that there was no history of trauma to the region.

Natal and post-natal history revealed that patient was delivered via naturalis at 32 weeks and six days of gestation. His birth weight was 2.1 kilograms and height was 10 inches. His father also revealed that patient had a large head and short limbs at the time of birth and had delayed milestones such as crawling, walking and speech.

The Extra-Oral features noted in this case were macrocephaly (head circumference was 56.8cm), disproportionately short stature, rhizomelic shortening of the arms and legs, limitations in joint movement, long face, frontal bossing, convex profile, midfacial hypoplasia, saddle nose, trident hands and polydactyl. Despite this dwarf like appearance, the child had normal intelligence. On intraoral examination mixed dentition was present in

the maxillary and mandibular arch, except for missing permanent maxillary left central incisor, and high frenal attachment were also observed. Multiple teeth were found to be carious, including a grossly destructed mandibular right first deciduous molar. Orthopantomograph normal skeletal structures and delayed eruption with respect to the maxillary left permanent central incisor and a chronic dentoalveolar abscess irt mandibular right first deciduous molar. Cephalometric analysis of the lateral skull radiograph revealed a posteriorly positioned maxilla relative to the nasion, a short cranial base, a normal mandible, a decreased upper facial height, an anteriorly tipped up palatal plane, and a skeletal class III jaw relation between the maxilla and mandible. Dentally, the maxillary incisors were proclined relative to the Frankfort horizontal plane, and the mandibular incisor angulations were normal relative to the mandibular plane.

A detailed genetic analysis revealed, the presence of heterozygous mutation in 1138 G > A loci of the Fibroblast Growth Factor Receptor 3 gene. Mutation in this gene causes a change in the sequence of amino acids from glycine to arginine, which in turn causes an inhibition in the growth of cartilage cells and disturbances in bone growth affecting the skull, spine and tubular bone.

As for the, missing maxillary left central incisor, it showed a normal eruption pattern in orthopantomograph. Hence, no treatment was planned for the same. Pulpectomy followed by stainless steel crown was planned for the mandibular right first deciduous molar.

Discussion

Achondroplasia is the most common form of dwarfism in humans. It occurs with a frequency of 1 in 15–25,000 and 80% of cases are sporadic. It is an autosomal dominant genetic disease that has 100% penetrance [1].

The achondroplasia phenotype has been recognized for thousands of years, as evidenced in the artifacts of many different cultures [2] and remains the most readily recognizable of the dwarfing disorders. The term seems to have been first used in the nineteenth century, and, while the main features were described shortly thereafter [3], it often was used as a generic descriptor of all short-limb dwarfing disorders. Detailed and specific radiologic and clinical features were carefully delineated by Langer et al. [4]. It remains the best characterized and most studied of the hundreds of dwarfing skeletal dysplasias.

Mutations in the FGFR3 gene cause achondroplasia. The FGFR3 gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. This specific mutation is at least 500-or 1000-fold more frequent than expected [5]. FGFR3 is one of four fibroblast

growth factor receptors in humans. All are cell surface receptors that influence cellular proliferation. FGFR3 is comprised of an extracellular domain with three immunoglobulin-like regions, a transmembrane domain and an intracellular tyrosine kinase [6].

Craniofacial Manifestations- can be delayed in achondroplastic children due to altered bone growth. Because of hypoplasia of midfacial structures malocclusion is common. In addition to maxillary hypoplasia, there is relative overgrowth of the mandible; it is uncertain whether mandibular growth is itself normal or diminished but less so than is the diminishment of maxillary growth [7]. These patients manifest midface hypoplasia and constricted maxilla with relatively large mandible which result in skeletal and dental class III, posterior crossbite, anterior reversed jet and anterior open bite. Early orthodontic evaluation should be considered in such children to attempt the possibility of interceptive orthodontics [8].

Management

Considerable progress has been made during the past twenty years in understanding FGFR3-related disorders as well in developing a rationale for effective therapeutic strategies to treat FGFR3-associated bone growth defects. Many nonsurgical strategies aimed at reducing excessive activation of FGFR3 **(Figure 1 and 2)** have been proposed to stimulate linear bone growth in patients with Achondroplasia, have been tried.

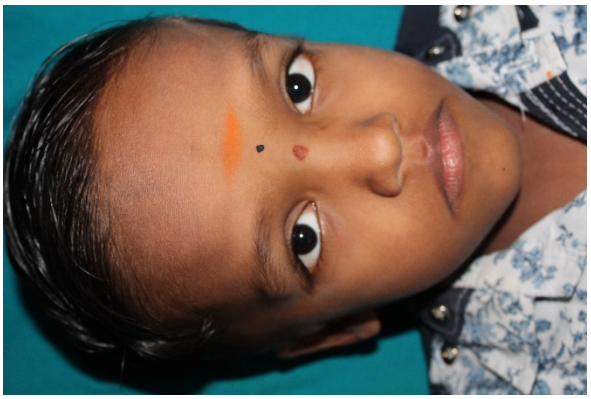

Figure 1. Brachycephalic skull, Frontal Bossing, depressed midfacial region, sunken nasal bridge.

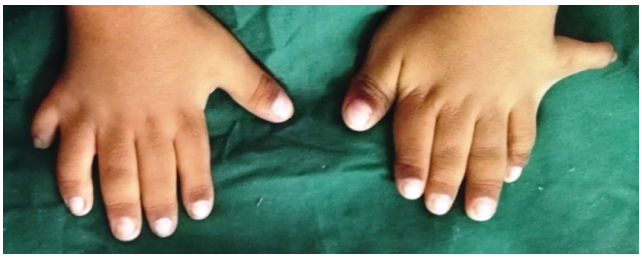

Figure 2. Rhizomelic Shortening of hands.

Conclusion

A great deal has been learned about the consequences of achondroplasia on those who are affected. Nonetheless, the quality of care that can be provided is compromised by the limited quality of the evidence that is, for the most part, available. Dentists treating these children should be able to recognize these features and its complications as dental management is constrained by practical problems associated with this disease.

References

1. Ornitz DM, Legeai-Mallet L (2017) Achondroplasia: Development, pathogenesis, and therapy. Dev Dyn 246: 291-309.
2. Pauli RM. (2019) Achondroplasia: a comprehensive clinical review. Orphanet Journal of Rare Diseases 14(1).
3. Baujat G, Legeai-Mallet L,Finidori G, Cormier-Daire V, Merrer ML (2008) Achondroplasia. Best Pract Res Clin Rheumatol 22:3–18.
4. Langer LO, Baumann PA, Gorlin RJ (1967) Achondroplasia. Am J Roentgenol 100:12–26.
5. Eswarakumar VP, Lax I, Schlessinger J (2005) Cellular signaling by fibroblast growth factor receptors. Cytokine Growth Factor Rev 16:139–49.
6. Goriely A, Wilkie AO (2012) Paternal age effect mutations and selfish spermatogonial selection: causes and consequences for human disease. Am J Hum Genet 90:175–200.
7. Shelmerdine SC, Brittain H, Arthurs OJ, Calder AD (2016) Achondroplasia: really rhizomelic?. Am J Med Genet A 170:2039–43.
8. Cardoso R, Ajzen S, Andriolo AR, deOliveira JX, Andriolo A (2012) Analysis of the cephalometric pattern of Brailian achondroplastic adult subjects. Dent Press J Orthod 17: 118–29.