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Achondroplasia – A Case Report

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Abstract

Achondroplasia is common autosomal dominant form of skeletal dysplasia and is caused by heterozygous mutation of the fibroblast growth factor receptor 3 (FGFR3) gene at 4p16.3 region. The phenotypic changes are mainly related to the inhibition of endochondral bone growth. It is an hereditary form of dwarfism, characterized by short stature and various clinical and radiographic features which are present in head, neck and face region. Hence oral physicians are in a position to diagnose such cases. The diagnosis is usually made on the basis of clinical findings and imaging features. In this paper, a case of 7-year-old girl child having clinical and radiographic features of achondroplasia, its management and modifications in dental treatment of such patient is discussed.

Keywords: Achondroplasia, FGFR3, Dwarfism, Trident hand configuration, Autosomal dominant

Introduction

The term Achondroplasia means without cartilage formation. It is a common form of chondrodysplasia, characterized by short limb dwarfism, affecting the growth of the tubular bones, spine and skull.[1] It is an autosomal dominant condition caused due to heterozygous mutation in Fibroblast Growth Factor Receptor-3 (FGFR-3) gene on 4p16.3 location which results in decreased endochondral ossification, inhibited proliferation of chondrocytes in growth plate cartilage, and decreased cartilage matrix[2]. Approximately 80% of cases are due to de novo dominant mutations.[3] The minimum is birth prevalence of 1.95 per 100000, found in south America while maximum birth prevalence of 52.25 per 100000 was found in north Africa and middle east. On an average, the worldwide birth prevalence is 4.6 per 100000.[9]

First use of term achondroplasia was done in 1878, to distinguish it from other bone abnormalities like rickets.[8] While cause of the condition i.e. FGFR3 mutation was discovered in 1995. As achondroplasia has autosomal dominant inheritance, both males and females ae equally affected. No documented race predilection is there.[8]

Characteristic clinical features of Achondroplasia are disproportionate short stature, macrocephaly, a prominent forehead (frontal bossing), hypoplastic middle third of face, rhizomelic shortening of limbs, trident hand configuration, prominent lumbar lordosis, and genu varum. Dentition is normal but most of the cases are reported with macroglossia.[2] We report a case of achondroplasia in a young girl child patient.

Case Report

A 7 year old girl child reported to department of oral medicine and radiology

with chief complaint of decayed teeth in her upper front region of jaw since 1 year. The patient had short stature and normal gait. No significant family history is there as her parents and elder sibling were normal. Her mother gives history of hearing impairment and recurrent ear infection in patient.

General physical examination showed short stature with rhizomelic shortening of limbs (Figure 1), trident configuration of hands with short and stubby fingers (Figure 6), prominent lumbar lordosis. The height of patient was 96 cm. Extraoral examination revealed macrocephaly, frontal bossing (Figure 2), midface hypoplasia with depressed nasal bridge resulting in concave facial profile and hypertelorism (Figure 3). While intraoral examination revealed multiple carious teeth and root stumps of deciduous teeth were present in chief complaint area with 61, 62 (Figure 4). Evident macroglossia were noted (Figure 5).

Figure 1. Short stature with Rhizomelic shortening of limbs

Figure 2. Midface hypoplasia with depressed nasal bridge and hypertelorism

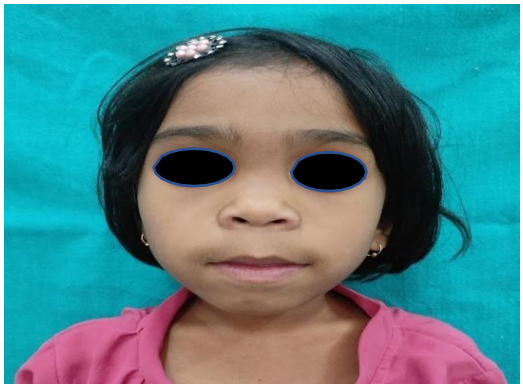

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Figure 3. Concave facial profile dueto midface hypoplasia

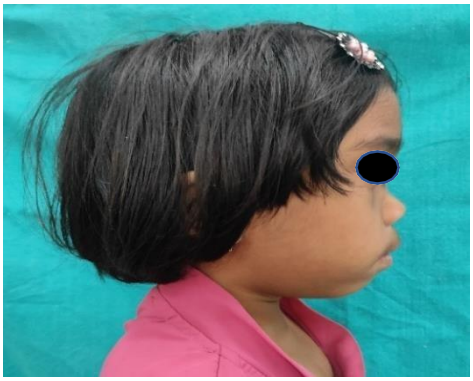


Figure 4. Chief complaint area, root stumps with 61,62

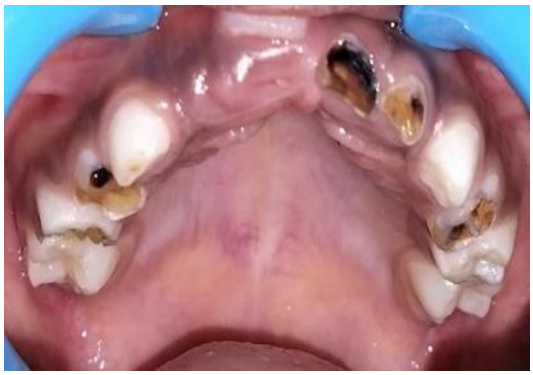


Figure 5. Macroglossia

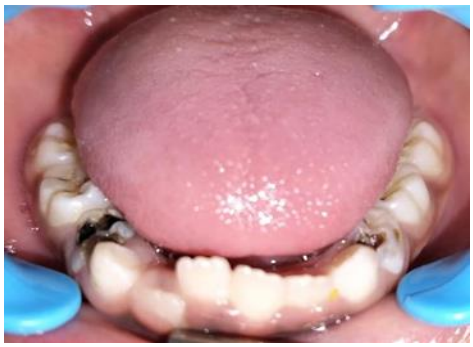
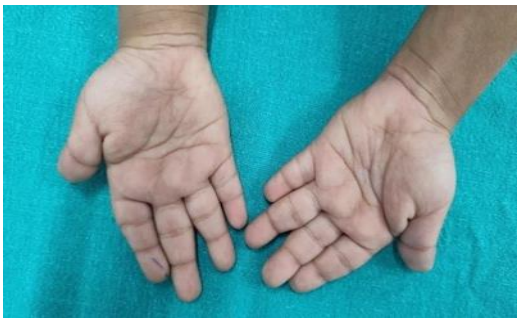


Figure 6. Trident hand configuration



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On radiographic examination her orthopantomogram showed impacted inverted maxillary left central incisor (Figure 7). Other developing permanent teeth were normal to her age. Lateral cephalogram revealed macrocephaly with relatively smaller skull base, evident frontal bossing was present (Figure 8). While hand-wrist radiograph showed small tubular bones i.e. short metacarpals and phalanges (Figure 9). Lateral abdominal radiograph showed prominent lumbar lordosis (Figure 10).

Figure 7. Orthopantomogram showing impacted inverted left maxillary central incisor

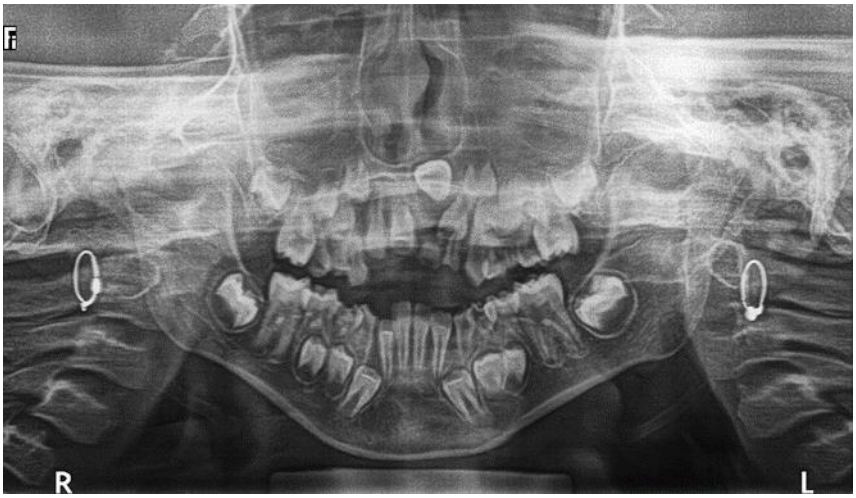

Figure 8. Lateral cephalogram Showing macrocephaly, Frontal bossing, small skull base.

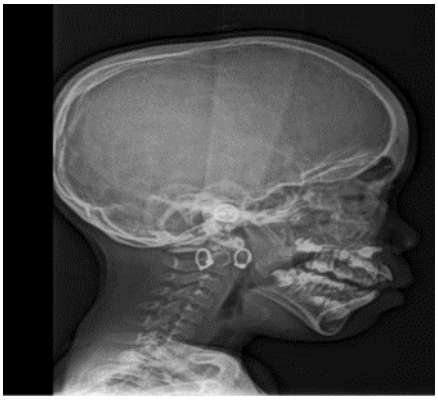

Figure 9. Hand-wrist radiograph showing short metacarpals and short phalanges.

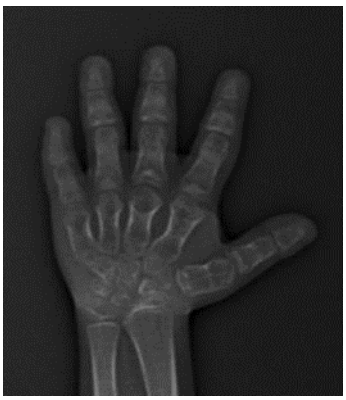

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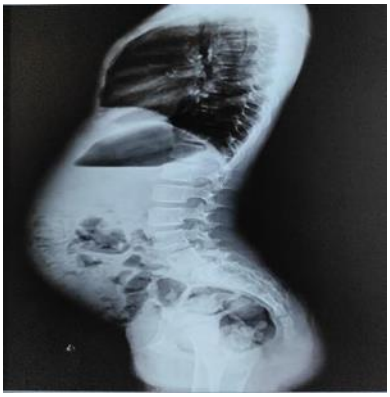
Figure 10. Lateral abdominal radiograph showing prominent lumbar lordosis



Discussion

Achondroplasia is an autosomal dominant disorder caused by mutation in FGFR3 gene on 4p16.3 region. Mutation of FGFR3 gene causes two other syndromes, they are hypochondroplasia and thanatophoric dysplasia. Hypochondroplasia is milder clinical form of achondroplasia in which trident hand configuration is not present.[4] Also, medical complications are rarely present. In thanatophoric dysplasia, severe thoracic and lung hypoplasia is present. It is lethal condition, most of infants dies in prenatal or early postnatal period.

Most cases are sporadic, means achondroplastic child can have normal parents. Over 80% of cases are present with spontaneous mutation.[5] In our case both the parents and her elder sibling were normal. So, it is a case of spontaneous mutation. Maximum height achieved in these patients, in males it is upto 130 cm and in females it is upto 123 cm.[2]

Many complications are present in achondroplasia like, foramen magnum compression (5–10%), craniocervical instability (very rare), symptomatic hydrocephalus (6%), recurrent otitis media (89%), and obstructive sleep apnea (75%).[3] Recurrent otitis media is the most common complication due to eustachian tube blockage, and over a period of time it may result into hearing loss. In our case, her mother reported mild hearing impairment. Obstructive sleep apnea arises because of a combination of midface hypoplasia resulting in smaller airway size and hypertrophy of the lymphatic ring.[1] In our case evident mouth breathing was present. Diagnosis is mainly based on clinical and radiologic criteria. Mutation detection is mainly useful for prenatal diagnosis.[5] There is currently no approved effective pharmacological treatment for skeletal dysplasias caused by variants of the FGFR3 gene.

Dentition of achondroplastic patients is usually normal, but most of cases are presents with dental crowding.[3] The previous cases reported in literature are also reported with skeletal and dental class III malocclusion, a narrow maxilla, macroglossia and an open bite between the posterior teeth.[2] During dental treatment, achondroplastic children may require special psychological management. The presence of disproportionate short stature, short limbs can cause a number of psychosocial and social problems along with dental problems like difficulty in sitting comfortably on dental chair, hence lowering the dental chair or using a step stool can be done in these cases. Furthermore, special precautions in head control during dental intervention are essential, due to the possible presence of hydrocephalus, craniocervical instability, foramen magnum stenosis and limited neck extension; as these may contribute to respiratory complications. Also, the presence of large tongue, large mandible increases difficulty of airway management in case of emergency.[7]

Conclusion

In this case report we presented the clinical and radiographic findings of a patient diagnosed with achondroplasia. This case highlights the importance of early recognition and diagnosis of achondroplasia allowing for appropriate management. Oral physicians are able to diagnose such cases because of presence of characteristic features in head, neck and face region.

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