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ACHONDROPLASIA – REPORT OF A RARE SYNDROME

Tulsi Subramaniam1, Arun Subramaniam*[2], Madhura Jathar[3] and Vaibhavi Raut[4]

[1]Professor, Dept. of Prosthodontics & Crown & Bridge, [2]Professor & Head, Dept. of Oral Medicine & Radiology, Dr. D.Y.Patil Dental College & Hospital, DPU, Pune, Maharashtra, India.
[3]Senior Lecturer, Sinhagad Dental College & Hospital, Pune, Maharashtra, India.
[4]PG student, Dept. of Oral Medicine & Radiology, Dr. D.Y.Patil Dental College & Hospital, DPU, Pune, Maharashtra, India.

ABSTRACT

Achondroplasia is rare anomaly which is heritable disorder of skeletal tissue growth. This syndrome is also known as 'chondrodystrophia fetalis'. It is considered as a very rare as its prevalence is significantly low. This disorder is characterized by dwarfism with marked disproportionate short limbs. A young man with achondroplasia reported typically with dwarfism, high forehead and various limb anomalies.

Key words: Achondroplasia, Dwarfism.

INTRODUCTION

Achondroplasia is autosomal dominant trait which brings defect in cartilaginous bone formation. It is a type of dwarfism with characteristics of short stature and short limbs. The term 'Achondroplasia' was first used in 1878 by Dr. Joseph Jules Marie Parrot (French physician). The prevalence of this disorder is 1 in 15000 (Anselmetti *et al*., 2011). Achondroplasia literally means failure to formation of endochondral bone formation. It occurs as a result of mutations in one copy of the fibroblast growth factor receptor 3 genes (FGFR3). More than 97% of patients have the same point mutation in FGFR3. (Ali Al Kaissi *et al*., 2008). The affected individuals may have disproportionate extremities, prominent forehead, dental malocclusion with anterior open bite. Apart from this physical deformity, the individual bears a normal intelligence level (Stephen *et al*., 2005).

Corresponding Author

Arun V Subramaniam
Email: arunsubramaniam66@gmail.com

CASE REPORT

A 21 year old male patient reported to outpatient, with the chief complaint of decayed tooth in lower left back region of jaw since 6 months. Patient also complained of lodgment of food in the same region. The patient was diagnosed as an achondroplasia case when he was a toddler. He gave no history of seeking medical treatment for his stunted growth. The patient presented classical features of achondroplasia. He was 122 cm in height. General physical examination showed large forehead (Figure 1), short, disproportionate limbs (Figure 2), comparatively larger head size and a stunted body growth. (Figure 3, 4).

On further questioning, there was no history of patient's parents consanguineous marriage. There was no relevant medical history given by him. Patient appeared to be well–adjusted, otherwise healthy and with normal intelligence level. The other developmental milestones were achieved at normal age. His vital signs were within normal range. On extra-oral examination, large forehead (figure 1), incompetent lips were noticed (Figure 7). Intra-oral examination revealed presence of 11, 13, 14, 15, 16, 17, 18, 21, 23, 24, 25, 26, 31, 32, 33, 34, 35, 37,38, 41, 42, 43, 44, 45, 46, 47 teeth. Both the arches were completely mal-aligned with an anterior open bite (Figure 6). The periodontium was otherwise healthy.

RADIOGRAPHIC FINDINGS

The radiographic investigation included lateral cephalogram, radiograph of thoraco lumbar region, orthopantomegraph and posteroanterior chest radiograph. The radiograph concerning to head region showed large cranium, frontal bossing and midfacial hypoplasia (Figure 8). The thoracolumbar region radiograph showed thoracolumbar kyphosis which is a typical feature of achondroplasia (Figure 9). This is also known as Gibbus deformity. The panoramic view showed the malocclusion of maxillary and mandibular arch, but the teeth were having normal size and shape (Figure 10).

On the basis of clinical features presented by the patient, history given by him and radiographic investigation, a confirmation of a case of achondroplasia was made.

Figure 1. Photograph showing relatively larger head and shorter limbs

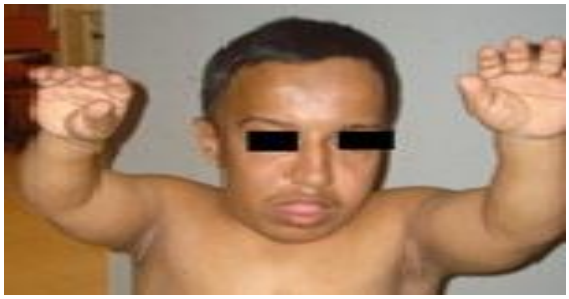


Figure 2. Short trunk

Figure 3 and 4. Deformed upper extremities

Figure 5. Intra oral photograph showing malaligned maxillary arch

Figure 6. Intra oral photograph showing malaligned mandibular arch

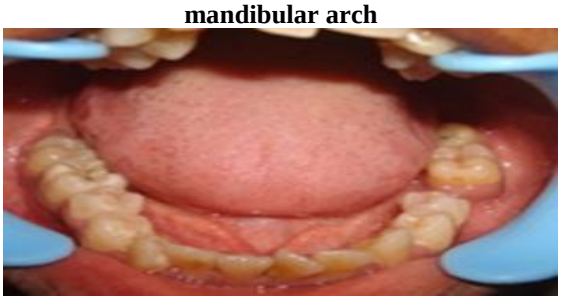


Figure 7.Clinical photograph showing anterior open bite

Figure 8. Photograph showing incompetent lips

Figure 9. Lateral view showing large cranium

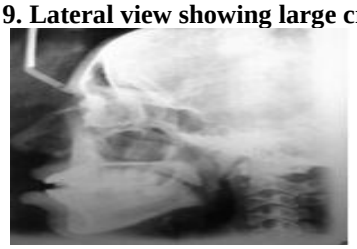


Figure 10. Thoraco lumbar kyphosis

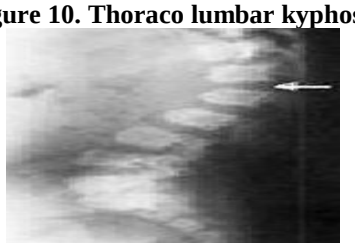


Figure 11. Malaligned upper and lower dental arches

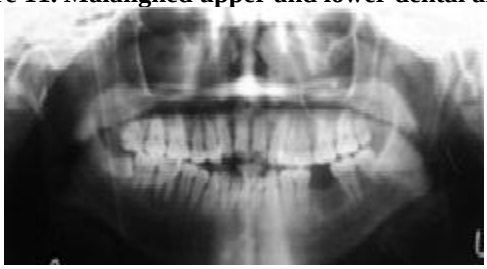
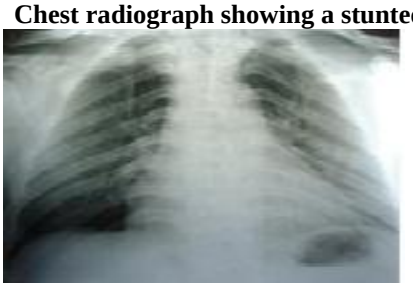


Figure 12. Chest radiograph showing a stunted rib cage



DISCUSSION

Achondroplasia is a non-lethal form of chondrodysplasia which means "without cartilage formation" (Chawla *et al*., 2012). Up to 90% of cases represent *de novo* mutations. It is inherited as autosomal dominant trait with complete penetrance (Kale *et al*., 2013). The common mutation causes a gain of function of the FGFR3 gene, resulting in decreased endochondral ossification, inhibited proliferation of chondrocytes in growth plate cartilage, decreased cellular hypertrophy and decreased cartilage matrix production; leading to a variety of manifestations and complications. The dentition in these patients is usually normal. In literature some cases of achondroplasia with delayed eruption are reported. (Staphne, 1950) reported retarded eruption of many permanent teeth in a 30-year-old affected male. Developmentally absent teeth have been reported rarely (Bellu *et al*., 1955). Most joints are hyperextensible, but extension is restricted at elbow (Sethi *et al*., 2011).

The mutation, which causes an increase in *FGFR3* function, affects many tissues, most strikingly the cartilaginous growth plate in the growing skeleton, leading to a variety of manifestations and complications (Crispian Scully, 2010).

The radiographic features of achondroplasia exhibit large calvarial bones with a small cranial base and facial bones. The vertebral pedicles are short throughout the spine as noted on a lateral radiograph. Hand radiographs show shortening of the metacarpals and phalanges with trident configuration (Shivapathasundharam, 2011; Ali Al Kaissi *et al*., 2008).

The differential diagnosis for achondroplasia includes hypochondroplasia, pituitary dwarfism, turner's syndrome, Leri Weill syndrome, constitutional delayed growth.

The quality of patient's life can be definitely improved by providing definitive dental management as well as medical attention. These patients require a psychological support to lead a normal life and to accept social challenges.

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