

# Achondroplasia: Case Report and Review of Literature

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## Abstract

A two month old male child presented to emergency of B. P. Koirala Institute of Health Sciences (BPKIHS), Dharan and was admitted to Paediatric ward with history of fever, cough and respiratory distress. On examination the breathing was rapid and shallow. The child was small for age with an upper to lower segment ratio of 1.9. The anteroposterior diameter of thorax was reduced. We report a very rare case of achondroplasia which was recognized in a two month age child who presented with severe pneumonia. Usually the clinically features of achondroplasia is more prominent when the child is growing in height but we diagnosed it in a two months child after performing the skeletal survey.

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## Introduction

Achondroplasia is autosomal dominant disorder and is the most frequent form of short-limb dwarfism. Affected individuals have rhizomelic shortening of limbs. Achondroplasia is caused by mutation in the fibroblast growth factor receptor-3 gene (FGFR3), which is located at 4p16.3. In Achondroplasia there are intrinsic disturbances in bone formation and modeling which causes disproportionate short stature and usually long bones of the body are affected because there is a defect in intracartilagenous ossification of the bone. The diagnosis is mainly based on clinical and radiological features in developing world and a mutation of gene causing achondroplasia can be detected by PCR<sup>1</sup>. Prenatal diagnosis can be performed by chorionic villous sampling at 10 weeks of gestation<sup>2</sup>. In achondroplasia and thanatophoric dwarfism, mutations are identified in fibroblast growth factor receptor 3 on chromosome 4. Achondroplasia become apparent only later in life as the child grows in height<sup>3</sup>. Although achondroplasia is a well documented cause of disproportionate short stature, its diagnosis in early infancy may be difficult because of the inherent disproportion of the child in the infancy as compared to older children or adults. Since achondroplasia is also associated with other problems, especially of the respiratory system, its early recognition is important to appropriately counsel the ignorant parents. We therefore report a two months

age child who presented with acute respiratory infection and diagnosed to be having achondroplasia based on clinical and radiological findings.

## The case

A two month old male child was admitted to pediatric ward of BPKIHS with a history of recurrent respiratory tract infections and respiratory distress. On examination the breathing pattern was rapid and shallow with associated abnormal breathing. The respiratory distress was in the form of nasal flaring and chest in drawing for which a diagnosis of severe pneumonia was made so the child was treated with standard protocol for treating severe pneumonia namely; cefotaxime, gentamycin, oxygen, intravenous fluid and nil per orally. The child was hospitalized for four days and the pneumonia resolved in three days and respiratory distress also resolved within that time. The child was nil orally for 40 hrs, intravenous fluid was given for 49 hrs and oxygen was required for 34 hours along with saline nebulisation. After treating the severe pneumonia and further on detailed examination it was found that there was a large cranium and the forehead was prominent. The nasal bridge was moderately flat, and the chest was small as compared to abdomen. The upper segment was bigger than the lower segment and these clinical manifestations made one to think about achondroplasia so a skeletal survey and anthropometric examination was further performed.

The skeletal survey showed features of achondroplasia. The anteroposterior diameter of thorax was reduced. The upper and lower segment ratio was 1.9. The head circumference was 40 cm, length was 47.5 cm and weight was 3.5 kg. X-ray of pelvis with bilateral lower limb anteroposterior view was performed which showed bilateral femora along with a short and wide tibiae with increased radiolucency at the upper end of femora and retarded ossification. Bilaterally iliac blades appeared small and square (Tombstone appearance). X-ray of the dorsolumbar spine anteroposterior and lateral view was also performed which revealed evidence of decrease in the interpedicular distance in the lower lumbar spine, suggestive of lumbar canal stenosis. X- Ray of skull lateral view was also performed which showed the bony calvarium large with a small sella. There was rhizomelic shortening of proximal end of long bones. All these findings were suggestive of achondroplasia.

Three distinct phenotype of achondroplasia exist, with group 1 possessing relative adenotonsillar hypertrophy, group 2 with muscular upper airway obstruction and progressive hydrocephalus and group 3 with upper airway obstruction without hydrocephalus. This child was kept in group 3 in which there was history of airway obstruction but there was no hydrocephalus.

## Discussion

The radiologic features of true achondroplasia and much concerning the natural history of the condition were presented by Langer et al (1967)<sup>4</sup> on the basis of a study of 101 cases and by Hall (1988). True megalencephaly occurs in achondroplasia and has been speculated to indicate effects of the gene other than those on the skeleton alone<sup>5</sup> Disproportion between the base of the skull and the brain results in internal hydrocephalus in some cases. In our case there was no hydrocephalus



**Fig 1:** Achondroplastic child with large head and rhizomelic (proximal) shortening of limbs.



**Fig 2:** X-ray skull lateral view showing large bony calvarium with small sella.



**Fig 3:** X-ray pelvis anteroposterior view with bilateral lower limb showing bilateral femora and tibiae appear short and wide with increased radiolucency at the upper end of femora. Bilaterally iliac blades appear small and square (Tombstone appearance).



**Fig 4:** X-ray dorsolumbar spine anteroposterior and lateral view showing decrease in the interpedicular distance in the lower lumbar spine suggestive of lumbar canal stenosis.

and monitoring of head circumference was static. The hydrocephalus may be caused by increased intracranial venous pressure due to stenosis of the sigmoid sinus at the level of the narrowed jugular foramina<sup>6</sup>. Hall et al (1988) pointed out that the large head of the achondroplastic fetus creates an increased risk of intracranial bleeding during delivery<sup>7</sup>. In present case there were no features of intracranial bleeds. Brainstem compression is common in achondroplasia and may account in part for the abnormal respiratory function<sup>8</sup>. Homozygosity for the achondroplasia gene results in a severe disorder of the skeleton with radiologic changes qualitatively somewhat different from those of the usual heterozygous achondroplasia; early death results from respiratory embarrassment from the small thoracic cage and neurologic deficit from hydrocephalus<sup>6</sup>. There is 2 reported female sibs as examples of achondroplasia; both died in the neonatal period and showed, in addition to chondrodystrophy, central harelip, hypoplastic lungs, and hydrocephalus<sup>9</sup>. In present case on auscultation we found wheeze but X-ray of chest revealed normal. In our case presentation the most common criteria for admission was severe pneumonia and the child improved with our standard protocol for treating severe pneumonia. The child was also kept under our zinc trial for treating severe pneumonia. As this trial is double blind placebo controlled study we are unaware whether the child got placebo or zinc.

## Conclusion

Although achondroplasia is well documented cause of disproportionate short stature, its diagnosis in early infancy may be difficult because of inherent disproportion of the child in infancy as compared to older children or adults. Achondroplasia is also associated with other problems; especially of the respiratory system. Early recognition becomes important to appropriately counsel the ignorant parents. As diagnosis of achondroplasia is difficult in infancy so careful history, clinical examination and skeletal survey are essential to detect achondroplasia in infants and early detection would help counsel the parents about the disease. Therefore careful history, clinical examination and skeletal surveys are essential for early recognition of achondroplasia.

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