# 2-Month-old Baby with Pneumonia and Accidentally Found "Champagne Glass Pelvic" Sign, Suspected Achondroplasia: A Case Report

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**Abstract**

**Background:** Achondroplasia is one of the most common types of skeletal dysplasia which causes short stature (dwarfism). Almost all types of achondroplasia are caused by unidentified genetic mutations in the fibroblast growth factor receptor type 3 (FGFR3) gene. Champagne Glass Pelvic appearance is a typical radiological sign of achondroplasia that occurs due to flattening of the iliac blades, increased acetabular angle and small sacroiciatic notch.

**Case Presentation:** A 2 month old baby was reported short of breath while undergoing treatment in the Pediatric Intensive Care Unit (PICU). Laboratory examination showed abnormal results with hematocrit 32.4 g/dL, platelets 585fl, CMPV 8.5 g/dL, Procalcitonin 0.500 vol%, segment 39.6 g/l, monocytes 14.5 vol%. On babygram radiology examination, there was an increase in bronchovascular pattern and homogeneous opacity in the bilateral lungs with an air bronchogram (+). The patient's heart had cardiomegaly with CTR>0.6. The ribs appear flat and narrow and the superior extremities appear bowed. The lower extremities appear bowed with a champagne glass appearance on the pelvic bones.

**Conclusion:** Various clinical and radiological findings can be found in someone who has achondroplasia. Genetic, clinical and radiological examinations are important to confirm the diagnosis of achondroplasia. The Champagne Glass Appearance radiological picture is very important in suspecting someone has achondroplasia.

Keywords: Achondroplasia, Babygram, Radiology, Imaging

## 1. Introduction

Achondroplasia (Ach) is one of the most common types of skeletal dysplasia which causes short stature (dwarfism). Even though radiological phenotypes and images have been studied for 50 years, they are still a medical problem that needs to be studied a lot today. The prevalence of this disease shows that there is 1 in 25,000 to 30,000 or 250,000 people experiencing achondroplasia. (Pauli RM, 2019). This disorder has been discovered for thousands of years and it has been identified that almost all types of achondroplasia are caused by unidentified genetic mutations in thefibroblast growth factor receptor type 3(FGFR3) gene (Shiang, et al, 1994).

The clinical features of achondroplasia generally include short stature, short limb and rhizomelic disproportion, macrocephaly, midfacial retrusion, small chest, thoracolumbar kyphosis, lumbar hyperlordosis, limited elbow extension, hypermobile hip and knees, and hypotonia. The radiological appearance of achondroplasia is generally squared off iliac wings, flat and horizontal acetabula, short proximal and middle phalanges (Pauli RM, 2019).

Champagne Glass Pelvic appearance is a typical radiological sign of achondroplasia that occurs due to flattening of the iliac blades, increased acetabular angle and small sacroiciatic notch (Roche,et al, 2002),

## 2. Case Reports

A 2 month old baby was reported short of breath while undergoing treatment in the Pediatric Intensive Care Unit (PICU). Laboratory examination showed abnormal results with hematocrit 32.4 g/dL, platelets 585fl, CMPV 8.5 g/dL, Procalcitonin 0.500 vol%, segment 39.6 g/l, monocytes 14.5 vol%. On babygram radiology examination, there was an increase in bronchovascular pattern and homogeneous opacity in the bilateral lungs with an air bronchogram (+). The patient's heart was found to have cardiomegaly with a CTR>0.6 so that the impression on the thorax was that there was bilateral pneumonia accompanied by cardiomegaly. The ribs appear flat and narrow and the superior extremities appear bowed. The lower extremities appear bowing with a champagne glass appearance on the pelvic bones, so it is suspected that this patient has achondroplasia.

## 3. Discussion

Longitudinal bone growth is influenced by processes called proliferation and differentiation of chondrocytes. The involvement of genetic mutations that occur on the short arm of chromosome 4 in the Achondroplasia gene accompanied by the substitution of arginine for glycine at residue 38 (p.Gly380Arg) in FGFR3 causes a direct relationship between FGFR3 and growth plate function. The mutant causes lysosome degradation which causes an increase in protein resulting in increased receptor signaling. FGFR3(p.Gly380Arg) mutation develops Ach-like phenotype with reduced chondrocyte proliferation (Ornitz DM,2017)

In this case, a 2 month old baby was found to be experiencing shortness of breath and was treated in the PICU. The patient underwent blood laboratory and radiological examinations. On blood laboratory examination, there were abnormalities in the form of hematocrit 32.4 g/dL, platelets 585fl, CMPV 8.5 g/dL, Procalcitonin 0.500 vol%, segment 39.6 g/l, monocytes 14.5 vol%. On radiological examination of the babygram, it was found on the thorax that there was increased bronchovascular patterning and homogeneous opacity in both lungs with an air bronchogram (Figure 1.A). The ribs appear flat and narrow with bowing at the superior extremities (figures 1.B and 1.C). In the lower extremities bowing and champagne glass appearance were visible (Figures 1.D and 2).

Several radiological images in previous studies explained that in neonates with achondroplasia the head circumference was more than 2 standard deviations from the average. Radiological images often show that the cranial vault membrane is disproportionately enlarged with frontal bossing and there is depression in the nasal area (Figures 3 and 4) (Glass, et al 2004). In the spinal section, patients with achondroplasia on radiological examination on Magnetic Resonances Imaging (MRI) can find posterior scalloping of the vertebrae as a result of a decrease in the interpediculate distance and width of the vertebrae which causes stenosis in the lumbar spine (Figure 5) (Wakely, 2006). Conventional photo examination in the lateral position can reveal kyphosis without secondary changes in the vertebrae at the top of the arch (Figure 6). The finding of a horizontal position of the sacrum as a result of hyperlordosis is often found in achondroplasia patients (Figure 7) (Pauli RM, 2019).

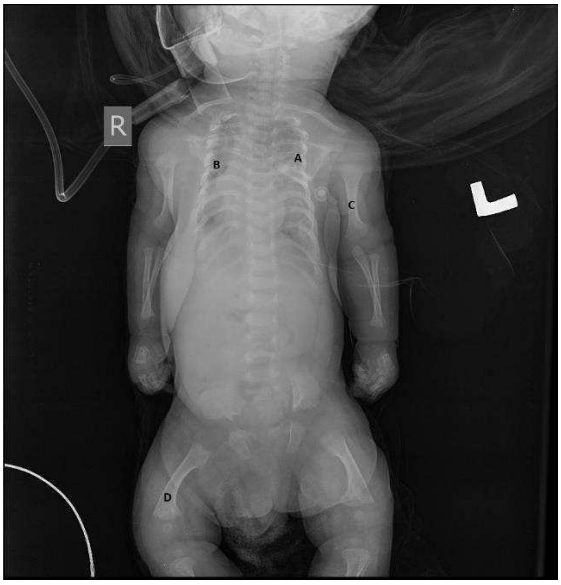


Fig.1(A) Increased bronchovascular pattern and homogeneous opacity in bilateral lungs accompanied by air bronchogram; (B)The ribs appear flat and narrow; (C) Bowing is seen in the superior extremities; (D) Bowing is seen in the lower extremities

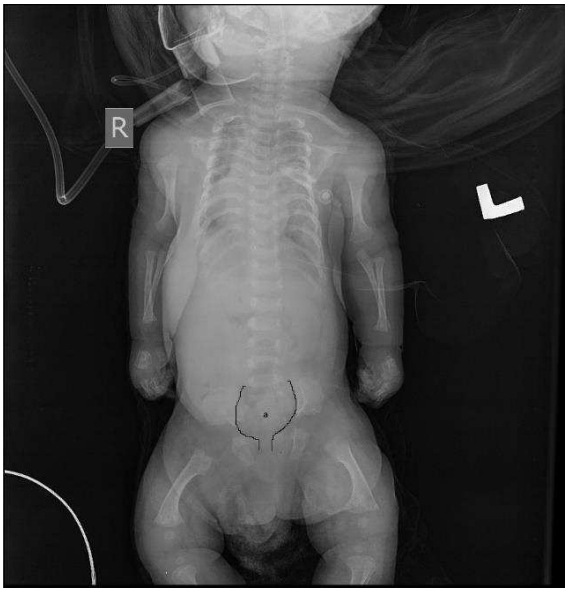


Fig.2. Imaginary line showing Champagne Glass Pelvic Appereance

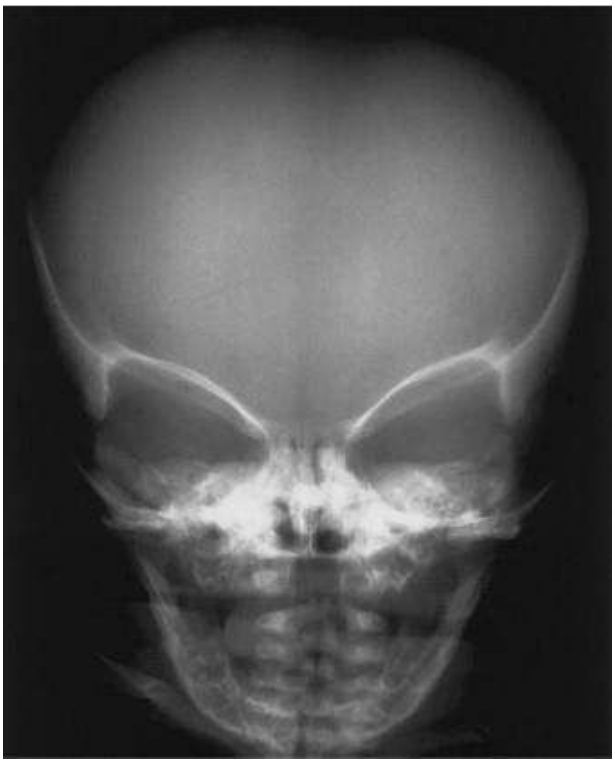

Fig.3. Frontal bossing shows that the vault membrane is disproportionately enlarged. (Original image taken from Pauli RM, 2019. Achondroplasia: a comprehensive clinical review. Orphanet Journal of Rare Disease.2019;14:1-49)

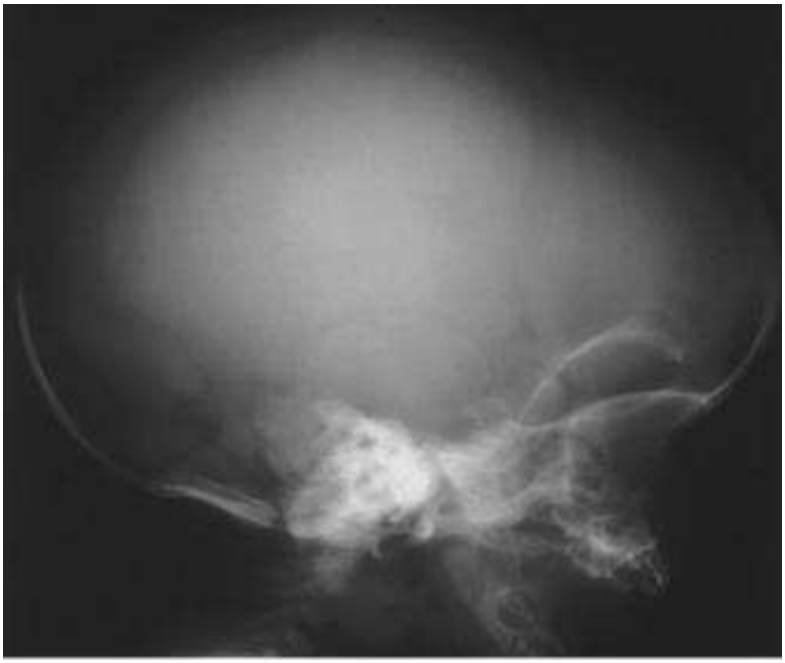

Fig.4. Image of Nosing depression in achondroplasia (Original image taken from Pauli RM, 2019. Achondroplasia: a comprehensive clinical review. Orphanet Journal of Rare Disease.14:1-49)

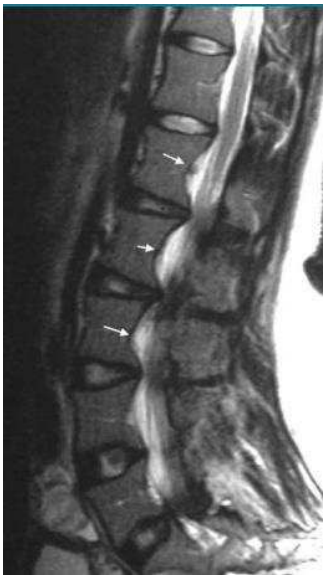

Fig.5. T2 MRI image of Posterior Vertebral Scalloping resulting from a decrease in the interpediculate distance and vertebral width which causes stenosis in the lumbar spine (Original image taken from Wakely, 2006: The Posterior Vertebral Scalloping Sign: Radiology.239(2):607-609)

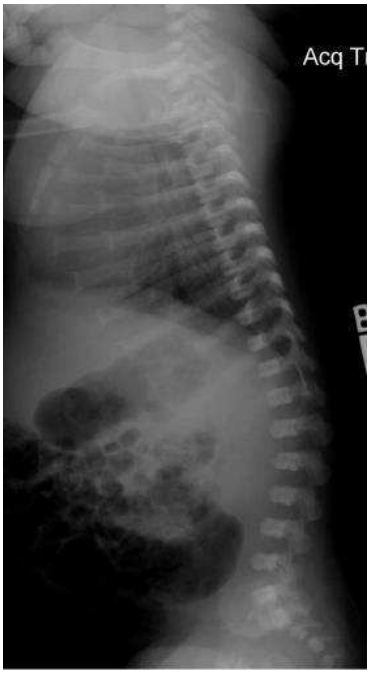


Fig.6. Image of kyphosis without secondary changes at the top of the arch (Original image taken from Pauli RM, 2019. Achondroplasia: a comprehensive clinical review. Orphanet Journal of Rare Disease.14:1-49)

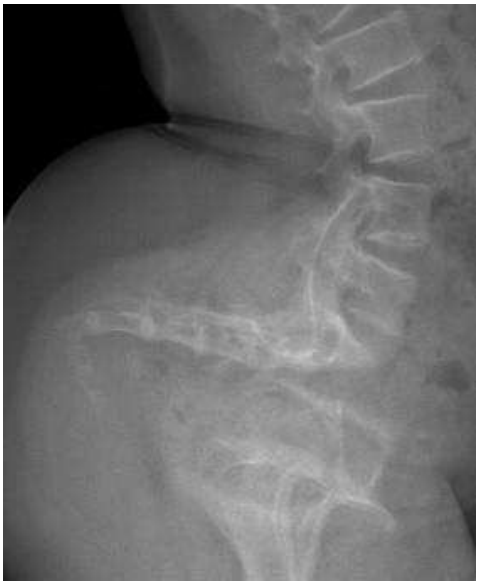

Fig.7. Image of a horizontal sacrum due to hyperlordosis (Original image taken from Pauli RM, 2019. Achondroplasia: a comprehensive clinical review. Orphanet Journal of Rare Disease.14:1-49)

## 4. Conclusion

Various clinical and radiological findings can be found in someone who has achondroplasia. Genetic, clinical and radiological examinations are important to confirm the diagnosis of achondroplasia. The Champagne Glass Appearance radiological picture is very important in suspecting someone has achondroplasia.

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