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# Prenatal Diagnosis and Multidisciplinary Management of Achondroplasia: A Case Report

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## CASE STUDY

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## Abstract

**Background**
Achondroplasia, the most common form of skeletal dysplasia, is characterized by rhizomelic limb shortening, macrocephaly, and distinctive facial features. Prenatal diagnosis relies on ultrasound findings and genetic testing, and the condition poses challenges due to its variable prognosis and significant clinical manifestations. This case report describes the prenatal diagnosis of achondroplasia at 22 weeks of gestation and discusses the clinical features, differential diagnosis, and multidisciplinary management.

**Case Presentation**
A 36-year-old woman, gravida 2 para 1, was referred at 22 weeks of gestation for evaluation after a second-trimester ultrasound revealed rhizomelic limb shortening, macrocephaly, and a narrow thoracic cavity. Amniocentesis confirmed heterozygous FGFR3 mutation diagnostic of achondroplasia. After multidisciplinary counseling, the parents opted to continue the pregnancy. Postnatal care was planned with a focus on respiratory support, orthopedic monitoring, and genetic counseling.

**Discussion**
Achondroplasia results from a mutation in the FGFR3 gene and follows an autosomal dominant inheritance pattern, often occurring de novo. Key differential diagnoses include hypochondroplasia and thanatophoric dysplasia. Multidisciplinary management is essential to address complications such as respiratory distress, spinal stenosis, and orthopedic deformities.

**Conclusion**
Early prenatal diagnosis of achondroplasia allows for informed parental decision-making and comprehensive postnatal care planning. A collaborative approach among obstetricians, geneticists, and pediatric subspecialists is critical for optimizing outcomes.

**Key Words**: Achondroplasia, Skeletal Dysplasia, FGFR3 Mutation, Prenatal Diagnosis, Rhizomelia.

## Introduction

Achondroplasia, meaning "without cartilage formation," is the most common form of skeletal dysplasia, affecting approximately 1 in 15,000 to 1 in 40,000 live births. It is caused by a mutation in the FGFR3 gene, which regulates endochondral ossification. The condition follows an autosomal dominant inheritance pattern, with approximately 80% of cases resulting from spontaneous de novo mutations, often associated with advanced paternal age [1].

Clinically, achondroplasia is characterized by disproportionate short stature, macrocephaly with frontal bossing, midface hypoplasia, rhizomelic limb shortening, and spinal abnormalities. Prenatal ultrasound findings, typically identified in the second trimester, include shortened long bones, macrocephaly, and thoracic hypoplasia. Genetic testing confirms the diagnosis, facilitating parental counseling and postnatal management planning [2].

This report describes a case of achondroplasia diagnosed prenatally at 22 weeks of gestation, highlighting the clinical findings, differential diagnoses, and the importance of a multidisciplinary approach to care.

## Case Présentation

The patient was a 36-year-old gravida 2 para 1 woman with no significant medical or surgical history. Her first pregnancy had resulted in a healthy child born at term via vaginal delivery. There was no family history of skeletal dysplasia or

congenital anomalies, and her marriage was non-consanguineous.

At 21 weeks of gestation, a second-trimester ultrasound revealed rhizomelic shortening of the long bones, macrocephaly, hypoplastic thoracic cavity, and short, broad extremities. Amniocentesis was performed, confirming a heterozygous FGFR3 mutation diagnostic of achondroplasia. No other abnormalities were detected in the fetal karyotype.

A 36-year-old gravida 2 para 1 woman was referred at 22 weeks of gestation for evaluation following an abnormal second-trimester ultrasound. She had no significant medical or surgical history, and her first pregnancy had resulted in a healthy child born at term. There was no known family history of skeletal dysplasia or congenital anomalies.

**Ultrasound Findings**

A detailed ultrasound examination revealed the following key findings:

1. **Rhizomelic Limb Shortening**: The femur length measured 1.32 cm, corresponding to less than the 2nd percentile for gestational age (**Figure 1**). The humerus length was also markedly reduced at 2.14 cm, consistent with disproportionate limb shortening.
2. **Macrocephaly**: The biparietal diameter (BPD) measured 5.84 cm, which was above the 86th percentile, with head circumference at the 45th percentile (**Figure 2**).
3. **Thoracic Hypoplasia**: A narrow thoracic cavity was evident, with chest circumference falling below the 6th percentile (**Figure 3**). This finding raises concerns about potential neonatal respiratory complications.
4. **Facial Features**: Ultrasound revealed frontal bossing and midface hypoplasia, typical craniofacial features of achondroplasia.

The estimated fetal weight was 237 g, consistent with gestational age, but the disproportion between the head and limb measurements strongly suggested a skeletal dysplasia. Amniocentesis was performed, confirming the diagnosis of heterozygous FGFR3 mutation associated with achondroplasia.

Multidisciplinary counseling was provided, and the parents opted to continue the pregnancy. Prenatal care included regular growth ultrasounds to monitor for complications such as polyhydramnios and further narrowing of the thoracic cavity. The delivery plan included anticipation of respiratory compromise and orthopedic deformities.

## Discussion

Achondroplasia, the most prevalent form of skeletal dysplasia, arises from mutations in the fibroblast growth factor receptor 3 (FGFR3) gene, leading to disproportionate short stature and characteristic craniofacial features. Early prenatal diagnosis is crucial for optimal management and parental counseling [3].

**Prenatal Diagnosis**

Ultrasound remains the cornerstone for detecting achondroplasia prenatally. Key sonographic markers include rhizomelic limb shortening, macrocephaly, and a narrowed thoracic cavity. These features typically become apparent in the second trimester, around 22 weeks of gestation. Studies have demonstrated that combining ultrasound findings with molecular genetic testing enhances diagnostic accuracy [4].

**Genetic Considerations**

Achondroplasia is an autosomal dominant disorder, with approximately 80% of cases resulting from de novo mutations. Advanced paternal age has been identified as a significant risk factor for these spontaneous mutations. Genetic counseling is essential to inform parents about recurrence risks and the implications of the diagnosis [5].

**Differential Diagnosis**

The prenatal presentation of achondroplasia can resemble other skeletal dysplasias, necessitating careful differentiation. Conditions such as hypochondroplasia, thanatophoric dysplasia, and osteogenesis imperfecta share overlapping features but differ in prognosis and management. Molecular testing aids in distinguishing these entities [6].

**Multidisciplinary Management**

A coordinated approach involving obstetricians, geneticists, neonatologists, and orthopedic specialists is vital for managing pregnancies complicated by achondroplasia. Prenatal planning should address potential complications, including respiratory distress due to thoracic hypoplasia and foramen magnum stenosis. Postnatal care focuses on monitoring growth, managing orthopedic issues, and providing developmental support [3].

**Parental Counseling**

Early and comprehensive counseling allows parents to make informed decisions regarding the pregnancy and prepares them for the anticipated challenges. Discussions should cover the spectrum of clinical manifestations, potential interventions, and long-term outcomes. Providing access to support groups and resources can also be beneficial [6].

**Advancements in Prenatal Testing**

The advent of non-invasive prenatal testing (NIPT) using cell-free fetal DNA has revolutionized the detection of genetic disorders, including achondroplasia. NIPT offers a safer alternative to invasive procedures like amniocentesis, with high sensitivity and specificity. However, its availability and cost may limit widespread use, particularly in resource-constrained settings [7].

## Conclusion

This case highlights the importance of prenatal diagnosis in skeletal dysplasias such as achondroplasia. Early

identification allows for comprehensive parental counseling and postnatal care planning. A multidisciplinary approach is essential to address the diverse complications associated with this condition and to optimize long-term outcomes. Future advancements in targeted therapies may provide new avenues for managing achondroplasia.

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## Figures

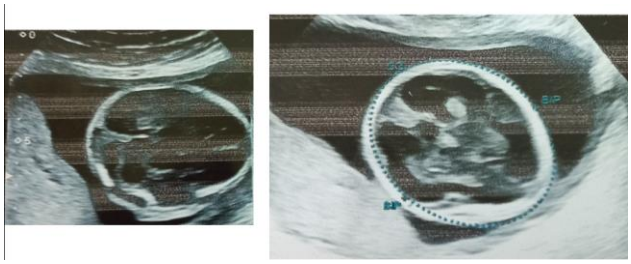

**Figure 1: Macrocephaly with a biparietal diameter of 5.84 cm and frontal bossing.**

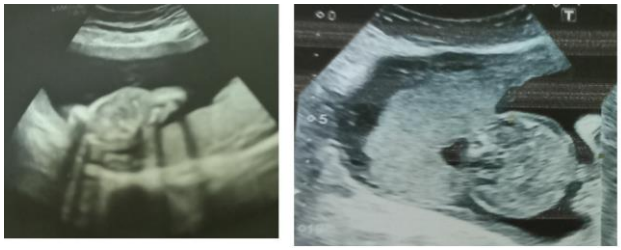

**Figure 2: Thoracic hypoplasia with reduced chest circumference.**

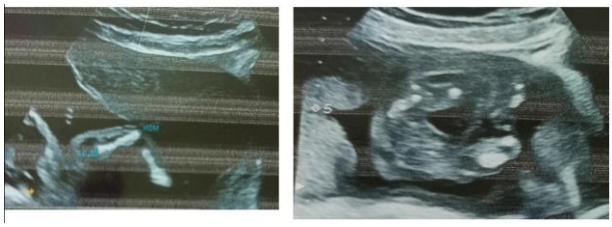

**Figure 3: Markedly shortened femur and humerus lengths (1.32 cm and 2.14 cm, respectively).**

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