International Journal of Research in Medical Sciences
*Arora R et al. Int J Res Med Sci. 2024 Aug;12(8):3027-3029*
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20241972

**Case Report**

# Thanatophoric skeletal dysplasia: a rare case report

**Rishabh Arora, Vijaya Narayana K. Holla***

Department of Radiodiagnosis, Srinivas Institute of Medical Science and Research Centre, Mukka, Mangalore, Karnataka, India

**Received:** 01 July 2024
**Revised:** 13 July 2024
**Accepted:** 15 July 2024

***Correspondence:**
Dr. Vijaya Narayana K. Holla,
E-mail: vijumagic@gmail.com



## ABSTRACT

A diverse range of bone growth disorders known as skeletal dysplasias are brought on by new mutations in the FGFR3 gene, which cause abnormalities in the size and shape of the skeleton. We present a case of thanatophoric dysplasia diagnosed antenatally on ultrasound. G2P1L0, a 30-year-old woman, came to our radiodiagnosis department for her first pregnant ultrasound examination. The results showed a single live intrauterine baby with a head that was considerably bigger than the rest of the body, a hypoplastic nasal bone, an exceedingly thin fetal thorax, thicker soft tissue in the hands and feet, as well as bent long bones that resemble telephone receiver handles, along with bilateral club feet and trident hands.

**Keywords:** Thanatophoric dysplasia, Congenital, Curved long bones

## INTRODUCTION

A diverse range of bone growth disorders known as skeletal dysplasias are brought on by new mutations in the FGFR3 gene, which cause abnormalities in the size and shape of the skeleton. Thanatophoric dysplasia is the most common form of skeletal dysplasia that is lethal in neonatal period.[1] Characteristics of thanatophoric dysplasia include severe shortening of the limbs, a narrow thorax, macrocephaly, and a normal trunk length.[1]

Clinically there are 2 recognized subtypes with some clinical overlap between the two subtypes. They can be differentiated by the skull shape and femur morphology: type 1 and type 2.

### *Type 1*

Characterized by micromelia with bowed femurs and, uncommonly, the presence of craniosynostosis of varying severity.

### *Type 2*

Characterized by micromelia with straight femurs and uniform presence of moderate-to-severe craniosynostosis with a cloverleaf skull deformity.[2]

This study aims to discover pertinent prenatal ultrasound imaging characteristics of this condition and to describe imaging features useful in differentiating between other skeletal dysplasias and thanatophoric dysplasia. Additionally, to find crucial prenatal ultrasound imaging features that will assist direct the care of the remaining pregnancy and advise the family on what to anticipate. We present a case of thanatophoric dysplasia diagnosed antenatally on ultrasonography.

## CASE REPORT

G2P1L0, a 30-year-old woman, came to our radiodiagnosis department for her first pregnant ultrasound examination. The Samsung HS70 machine's curvilinear

probe transducer was used at our institution to do ultrasonography.

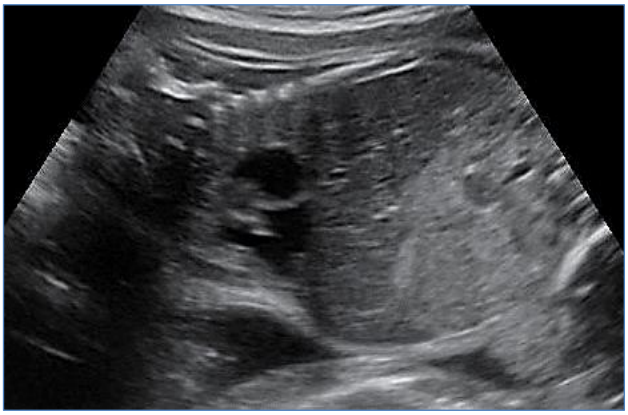

**Figure 1: Extremely narrowed fetal thorax.**

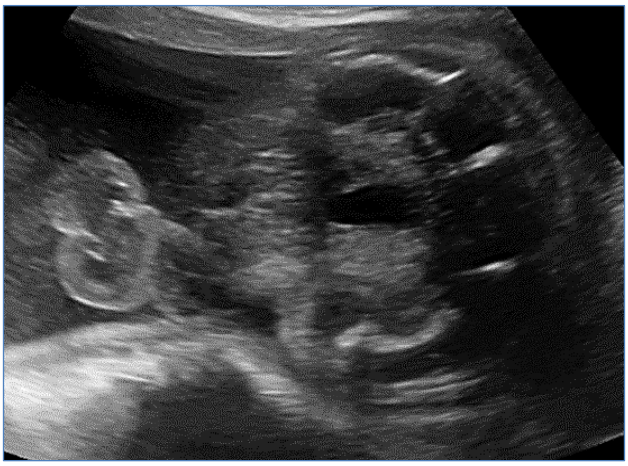

**Figure 2: Short, thick, bowed tubular femur having a typical "telephone handle" bowing with metaphyseal flaring.**

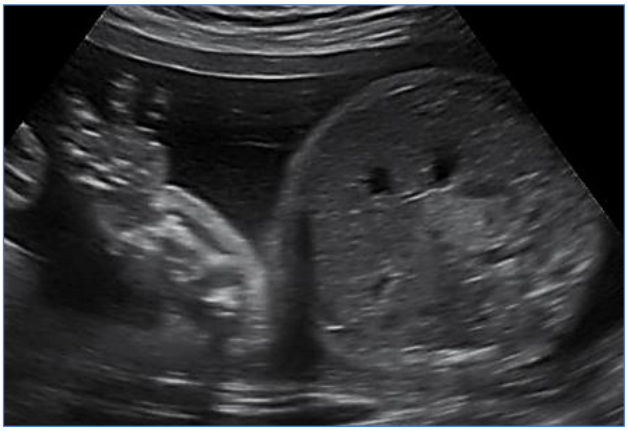

**Figure 3: Short and thick radius and ulna.**

The results showed a single live intrauterine baby with a head that was considerably bigger than the rest of the body, a hypoplastic nasal bone, an exceedingly thin fetal thorax, and nasal bridge flattening. Evidence of pulmonary hypoplasia is indicated by a small chest, a reduced thoraco/abdominal ratio, and hypoplastic both lungs. There is evidence of thicker soft tissue in the hands and feet, as well as bent long bones that resemble telephone receiver handles, along with bilateral club feet and trident hands. Femur was 18 mm (15 days and 4 weeks), humerus: 19 mm (16 days and weeks), and tibia: 17 mm (15 days and 3 weeks). 18 mm is the radius (17 weeks 0 days). Fetal bone mineralization was found to be adequate.

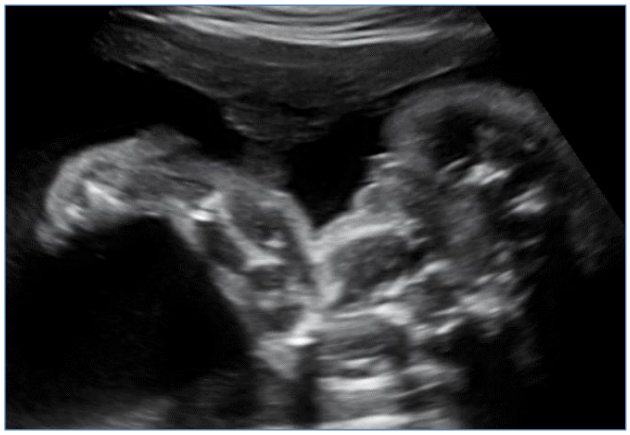

**Figure 4: Short and thick lower limb bones with thickened soft tissues and associated club foot.**

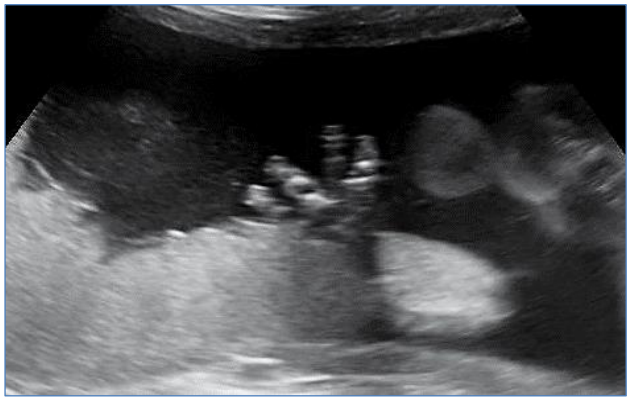

**Figure 5: Hand with short and stubby fingers, with a separation between the middle and ring fingers, suggestive of trident hand.**

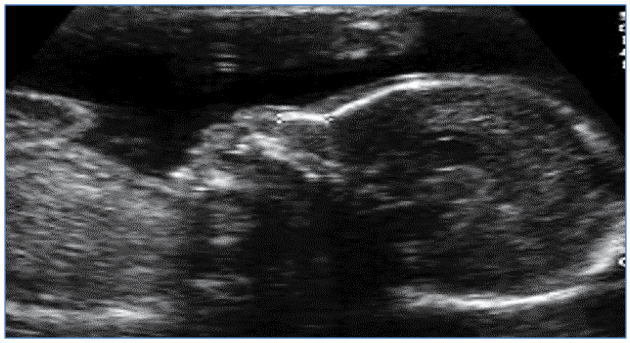

**Figure 6: Hypoplastic nasal bone (2.4 mm).**

**Figure 7: Post abortal image-consistent with antenatal ultrasonographic findings and suggested diagnosis of thanatophoric dysplasia.**

## DISCUSSION

Thanatophoric dysplasia is characterized by distinct skeletal morphology and anthropometry. The mode of inheritance is a new autosomal dominant mutation. The existence of a femur resembling a curved telephone handle in our instance verified type 1 TD, but type 2 TD was ruled out in both situations due to the lack of a cloverleaf head. Clinical parallels have, however, only very seldom been documented.

Prenatal diagnosis can be made by ultrasound in the second or the third trimester. Ultrasound findings are generalized micromelia, with short curved or straight femora, large or cloverleaf head, small narrow chest, small hands and feet, and flat hypoplastic vertebral bodies. Radiological examination reveals a disproportionate large skull, a very narrow thorax with shortened, cupped ribs, severe platyspondyly, and generalized but preferential rhizomelic dwarfism. The characteristic "French telephone receiver" aspect of the femora and humeri is typical for the type 1 TD. The long bones are straight in type 2 TD. The vertebrae are H-shaped on the AP-view, due to a decrease in the height of the vertebral bodies, with a sparing of the height of the pedicles.[3]

The differential diagnoses of thanatophoric dysplasia include: achondroplasia (occurs in both parents) is one of the differential diagnoses for thanatophoric dysplasia; chondrogenesis (shortened trunk length; most pronounced bone demineralization in the calvarium and vertebral bodies); campomelic dwarfism (long bones bowed and angulated due to immature ossification); phizomelic chondrodysplasia punctate which is characterized by rhizomelic micromelia that have radiological stippling and punctuate calcification in the cartilage; and severe hypophosphatasia and severe imperfect osteogenesis (widespread hypomineralization of bones resulting from many fractures).

## CONCLUSION

A comprehensive and expert prenatal sonographic fetal diagnosis is necessary for early family counseling in the case of thanatophoric dysplasia, a rare inherited skeletal abnormality. This condition is unique clinically and radiologically. The diagnosis of thanatophoric dysplasia is strongly suggested by ultrasonography; however, confirmation is obtained by prenatal genetic investigation, clinical characteristics at birth, or postmortem. In utero, most fetuses perish away. The cause of death was respiratory insufficiency, which might have resulted from a combination of the brain stem compression caused by a short foramen magnum and hypoplastic lungs, or from a limited chest cavity alone.

*Funding: No funding sources*
*Conflict of interest: None declared*
*Ethical approval: Not required*

## REFERENCES

1. Sahu S, Kaur P. Thanatophoric Dysplasia: Antenatal Diagnosis. Med J Armed Forces India. 2009;65(1):87-8.
2. Handa A, Nishimura G, Zhan MX, Bennett DL, El-Khoury GY. A primer on skeletal dysplasias. Jpn J Radiol. 2022;40(3):245-61.
3. Handa A, Grigelioniene G, Nishimura G. Skeletal Dysplasia Families: A stepwise Approach to Diagnosis. Radiographics. 2023;43(5):e220067.

**Cite this article as:** Arora R, Holla VNK. Thanatophoric skeletal dysplasia: a rare case report. Int J Res Med Sci 2024;12:3027-9.