# Prenatal Third Trimester Sonographic Behavior of a Thanatophoric Dwarfs

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**Summary**

***Background.*** **Thanatophoric dysplasia (TD), the most common of the congenital lethal skeletal dysplasias occurs sporadically in 1/64,000-100,000 live births. To the best of our knowledge, the in utero attitudes and behaviours of such babies with serial ultrasound scans have not been previously described.**
***Objectives.*** **To present the in utero third trimester sonographic behaviors of TD in a 22-year-old primigravida diagnosed at '32weeks' gestational age along with the clinical and radiographic characteristics.**
***Methods.*** **The same radiologists to observe the behavior of a thanatophoric dwarfs did three fortnights serial ultrasound scans.**
***Results.*** **The baby was found to have short limbs that were constantly in rigid abduction, flexed at both elbows and knees, and demonstrated poverty of synchronous movements. The upper limbs were perpetually in embracing position during all scans. In addition, he was hyperactive, showing "yoyo" body movement and constantly hyper-extended neck. Postmortem radiograph was diagnostic of TD.**
***Conclusion.*** **Though he baby died intra partum, the observed attitudes and behaviors on serial prenatal ultrasonography, which provided us with sufficient information to counsel the family, managed the pregnancy, and direct the postnatal evaluation could possibly add to the in utero diagnostic sonographic features of TD.**

*KEY WORDS: dwarfism, fetal anomalies, obstetrics ultrasonography, plane radiograph, thanatophoric dysplasia.*

## Introduction

Thanatophoric Dysplasia/Dwarfism (TD) is a name derived from a Greek word 'Thanatophores' which means constantly bearing death. It was first described by Maroteaux et al in 1967 (1). This rare congenital anomaly occurs sporadically in 1/64,000-100,000 total live births and is the most common of the congenital lethal skeletal dysplasias (1-7). It is characterized by short limbs, small conical thorax, platyspondyly and macrocephaly (1-7), and the affected fetuses usually die within the first 48 hours of life (1-4).
There are two variants (1-5); The type 1 variant presents with polyhydramnios, macrocephaly, short limbs, narrow thoracic cage and curved short femur (the typical telephone receiver appearance), but without a cloverleaf skull. On the other hand, short limbs, narrow thoracic cage, straight short femora, hydrocephalus, and cloverleaf skull (1, 7) characterize the type 2 variant. These sonographic and radiographic features are the diagnostic characteristics that differentiate TD from achondroplasia and achondrogenesis. Although TD can be distinguished from other short-limbed dysplasia syndromes with characteristics prenatal sonographic features, the prenatal ultrasound diagnosis can be difficult (1-7). The definitive diagnosis of TD is often made postnatal based on clinical and radiographic criteria.
While the phenotypic, sonographic and radiographic features of TD have been extensively described in the literature, there is dearth of information on the in utero attitudes of thanatophoric baby in the literature. Therefore, we present the in utero third trimester behavior of a case in a 22-year-old primigravida diagnosed at '32weeks' gestational age on prenatal ultrasound followed up to delivery by serial ultrasound scans together with the clinical, sonographic and radiographic characteristics.

## Case report

A 22-year-old primigravida was referred from a private clinic for routine obstetrics scan at our unit. She is learned and was sure of her last menstrual date. Her symphysofundal height was non-corresponding to her date (36cm to 30weeks). At sonography, there was a single live intra uterine male fetus in longitudinal lie with cephalic presentation. The limbs were short, constantly in rigid abduction, flexed at both elbows and knees, and demonstrated poverty of synchronous movements. The upper limbs were perpetually held in an embracing position during all scans done at different days (Fig. 1A-C). The fetus was hyperactive showing rhythmic segmentation body movement that could be likened to a "yoyo". The fetal neck was hyper-extended in all series of scans.

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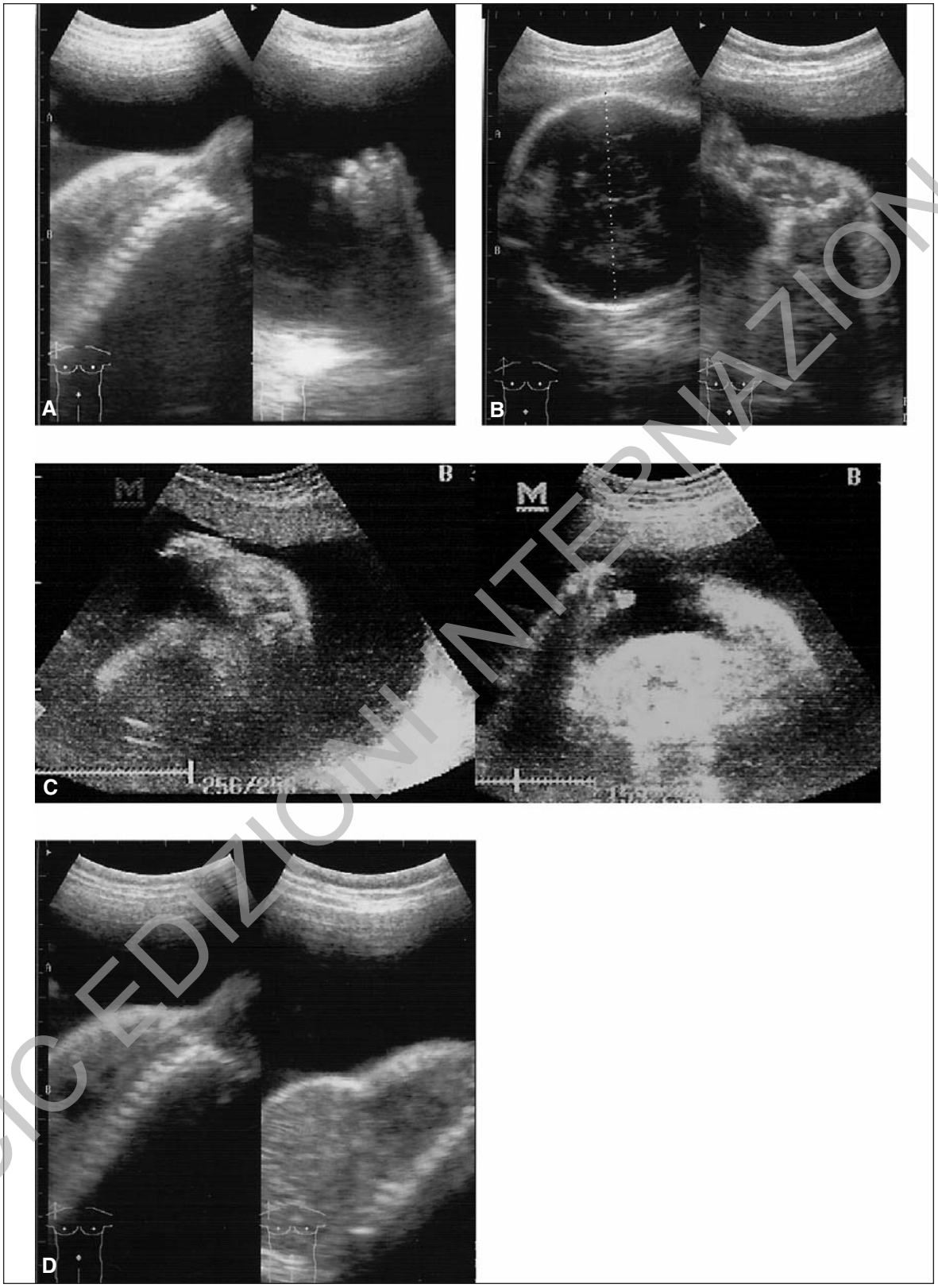


Figure 1 - Prenatal sonograms of a thanatotrophic dwarfs. (A) Demonstrate fetal trunk (right) and fetal hand (left) at 32weeks gestational age; (B) demonstrated fetal skull and the short lower limb; (C) Shows both upper limbs of the fetus in persistent embracing position, and (D) Shows narrowed fetal trunk and the protuberous abdomen fetal trunk. Note the persistence of fetal neck in extension, the widened intervertebral spaces and tongue-like projections of the vertebral bodies anteriorly in (A) and (D).

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The narrowed thorax and the protuberous abdomen gave a dumbbell appearance (Fig. 1D). The femora were barely demonstrable. When both tibia and fibula lengths was 10.8mm (under range for Gestational age assessment), the biparietal diameter was 94.2cm (equivalent to 38weeks 3day gestational age). The brain parenchyma and the ventricular system showed normal sonographic appearances. The spine showed flattened vertebral bodies that tongued out anteriorly and wide intervertebral spaces (Fig. 1A & D). Placenta was anterofundal and appears normal. There was polyhydramnios. A diagnosis of multiple skeletal dysplasia with strong suspicion of TD was made. The parents were counseled on the possible outcome and management options. However, both parents agreed to carry the pregnancy to term and wait for the natural outcome rather than the option of termination. At the end of 39 weeks, she went into spontaneous labour and delivered a fresh lifeless boy that weighed 2.8 kg. The body length was 34 cm and the head circumference was 39cm. The calvarium and fontanelles were slightly enlarged. The nasal bridge was depressed and the ears were low set. He had thick and protuberant tongue (Fig. 2A). The extremities were very short with each arm and thigh measuring 7cm and 6cm respectively in length, and the legs 8cm in length. The digits were short and thick. Many deep skin folds were noted in the extremities (Fig. 2A). The thorax was narrowed and the abdomen was protuberous.

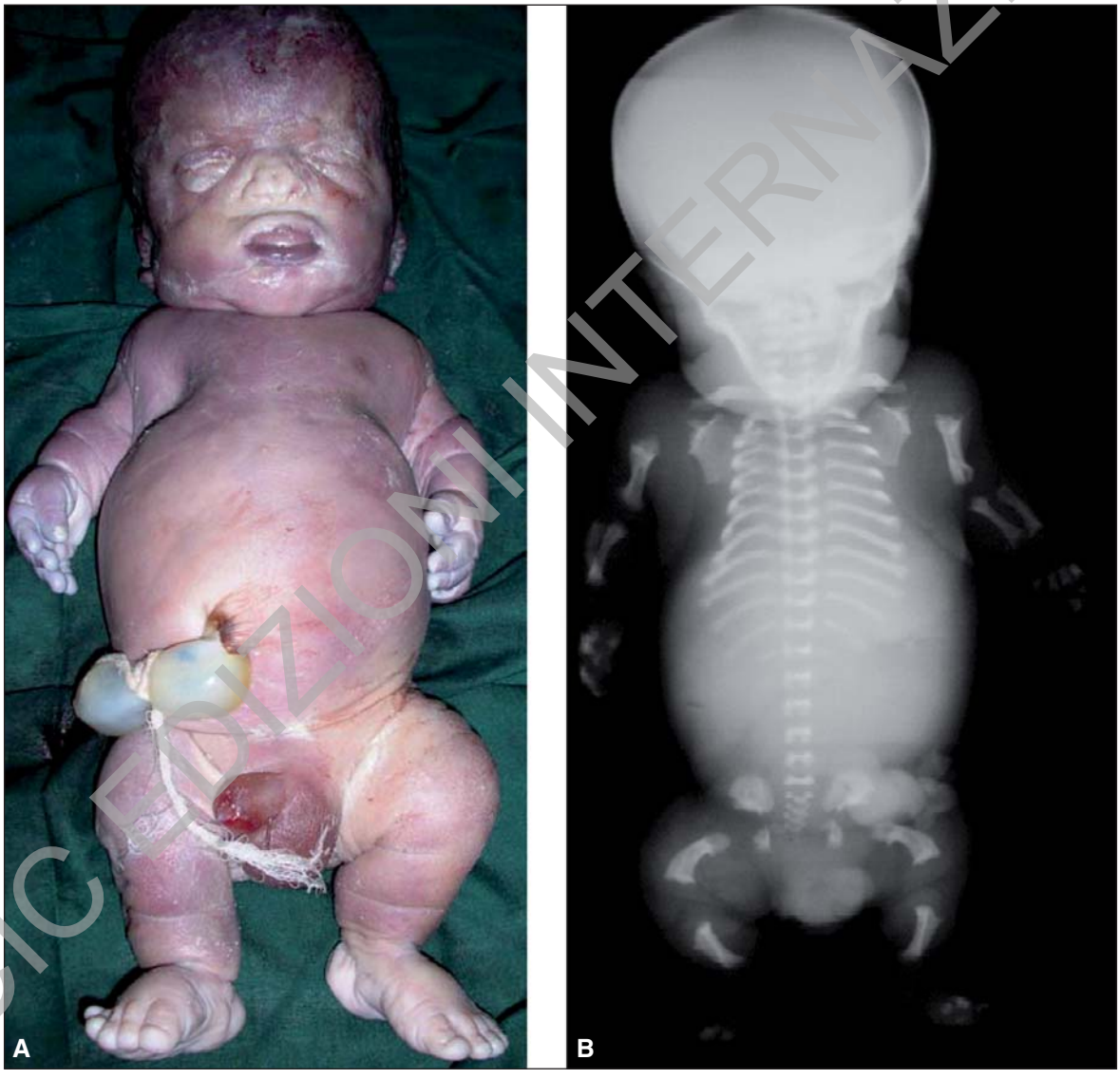


Figure 2 - Photography and postmortem Diagnostic babygram of the thanatophoric dwarfs at birth. In (A), note the depressed nasal bridge, low-set ears, thick and protuberant tongue, very short limbs, short and thick digist and the many deep skin folds in the extremities. Note also the narrow thorax and the protuberant abdomen. (B) Postmortem radiograph demonstrating narrowed thorax, short ribs with bead-like thickening anteriorly, hypoplastic lungs, short and small scapulae. The facial bones are small compared to the calvarium, the nasal bridge is depressed, the tubular bones of the arms and legs are short and moderately bowed to give telephone handle appearance, metaphyses are irregularly flared, vertebral bodies were U-shaped and the iliac bones have a greater width than height and a horizontal inferior border with flattened acetabulae.

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The postmortem radiograph demonstrated narrowed thorax and short ribs with bead-like thickening anteriorly. The lungs were unexpanded (hypoplastic). The scapulae were small and short. The clavicles were normal. The facial bones were small compared to the calvarium and the nasal bridge was depressed. The tubular bones of the upper and lower limbs were extremely short, showed irregular flared metaphyses and moderate bowing that gave "telephone handle" appearance. The vertebral bodies were U-shaped. The iliac bones had greater width than height, horizontal inferior border and flattened acetabulae. These features characterized thanatophoric dwarfism. However, religion and cultural beliefs were hindrance to an autopsy examination.

## Discussion

Thanatophoric dwarfism (TD) is the most common lethal skeletal dysplasia, with an unknown aetiology. It is commonly mistaken clinically for achondroplasia, particularly the heterozygous type in which both parents are of normal stature (1-7). This confusion is unlikely when one or both parents are achondroplastic dwarfs, since a TD offspring from this combination has not been reported (1, 3). All reported cases of TD have been in singleton pregnancies except for one case, the first known case of one fraternal triplet affected by thanatophoric dwarfism reported by Oga et al. (8). The only female infant of this triplet diagnosed at 18 weeks gestation and delivered at 30 weeks gestation by cesarean section suffered TD.
Several publications and review have described the clinical, sonographic and radiographic features of TD (1-10), but none described the in utero attitude of such babies with serial scans as presented in this case. Flattened vertebral bodies with widened intervertebral spaces and shortened-bowed tubular bones of the extremities with irregularly flared metaphyses represent the most important radiographic features differentiating TD from other short-limb dwarfisms, particularly achondroplasia.
We agreed with Spirt et al. (2) that prenatal sonographic evaluation of short-limbed dwarfism should be initiated when a significantly shortened femur is found. To differentiate the type of bone dysplasia and determine whether it is lethal; the fetal spine, head, thorax, hands, and feet should be carefully evaluated. In such instance, all the long bones should be measured and evaluated for bowing, fractures, and densities (1-6). Dwarfism with reduced bone densities and multiple fractures are characteristic in ostegenesis imperfecta as dwarfism with bowing and metaphyseal irregularities as it is in our case should suspect TD rather than achondroplasia. Serial prenatal ultrasound examinations as done in our case may provide sufficient information to diagnose, counsel the family, manage the pregnancy, and direct the postnatal evaluation. The observed fetal attitude and behaviors: hyper-extended neck, upper limbs in persistent embracing position, and rhythmic segmentation trunk movement likened to "yoyo" observed in this case could possibly add to the in utero diagnostic sonographic features of TD.
Radiological imaging plays a major role in making an accurate diagnosis, but despite recent advances in imaging, fetal skeletal dysplasias are difficult to diagnose in utero due to a number of factors (1-7). These include: the large number of skeletal dysplasias and their phenotypic variability with overlapping features; lack of precise molecular diagnosis for many disorders; lack of a systematic approach, the inability of ultrasonography (US) to provide an integrated view; and variability in the time at which findings manifest in some skeletal dysplasias. US of suspected skeletal dysplasia require systematic imaging of the long bones, thorax, hands and feet, skull, spine, and pelvis.
Assessment of the fetus with three-dimensional US has been shown to improve diagnostic accuracy, since additional phenotypic features not detectable at two-dimensional US may be identified (4, 9). Obstetric ultrasound examination is a primary diagnostic tool in the antenatal detection of TD. US examination is capable of diagnosing TD from second trimester to delivery and detecting other associated abnormalities, including polyhydramnios, hydrops, and CNS abnormalities, particularly abnormal temporal lobe development (7). The obstetric sonographic examination alone, in certain cases, may be sufficient to make a definitive diagnosis, but when combined with radiography, the definitive diagnosis of TD is certain (5). Recently, prenatal ultrasonography in combination with the molecular confirmation of fibroblast growth factor receptor 3 gene mutation have been helpful in early diagnosis and genetic counseling of thanatophoric dysplasia (9, 10).
According to Spirt et al. (2) who presented an organized approach to the detection and evaluation of bone dysplasias in utero, the main objectives of prenatal detection are to counsel the family, manage the pregnancy, and direct the appropriate postnatal radiologic evaluation. Postnatal examination and detailed radiographic examination of the fetus (Baby-gram) are essential to precisely identify the type of skeletal dysplasia, which is paramount for proper genetic counseling (6).
The prognosis of TD is bad. In a retrospective sonographic evaluation of 13 short-limbed fetuses with lethal skeletal dysplasias composing of thanatophoric dwarfism, achondrogenesis, osteogenesis imperfecta and campomelic dwarfism by Pretorius et al. (3), deaths occurred in utero or within 2 weeks after delivery in all cases. Where the affected parent consented, pregnancy termination can be offered. In our case, both parents agreed to carry the pregnancy to term and wait for the natural outcome rather than the option of termination. However, the baby died intra partum.

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