World Journal of Advanced Research and Reviews
eISSN: 2581-9615 CODEN (USA): WJARAI
Cross Ref DOI: 10.30574/wjarr
Journal homepage: https://wjarr.com/

(CASE REPORT)

Prenatal diagnosis of thanatophoric dysplasia: A case report

Younes Tazi Moukha *, Hikmat Chaara, Yassine belhaj, Sofia Jayi, Fatima Zohra Fdili Alaoui and Moulay Abdelilah Melhouf

Department of Obstetrics and Gynecology II, Hassan II University Hospital, Fez, Morocco.

World Journal of Advanced Research and Reviews, 2026, 29(03), 736-740

Publication history: Received on 27 January 2026; revised on 08 March 2026; accepted on 11 March 2026

Article DOI: https://doi.org/10.30574/wjarr.2026.29.3.0553

Abstract

Thanatophoric dysplasia (TD) is a rare and uniformly lethal skeletal dysplasia caused by activating mutations in the FGFR3 gene located on chromosome 4p16.3. It represents one of the most severe forms of osteochondrodysplasia diagnosed in the prenatal period. Although molecular genetic testing confirms the diagnosis, characteristic imaging findings frequently allow reliable prenatal identification. Two phenotypic subtypes are described: type I, characterized by curved long bones, and type II, associated with a cloverleaf skull deformity.

We report the case of a 26-year-old nulliparous woman with an unsupervised pregnancy who presented at 34 weeks of gestation with threatened preterm labor. Ultrasound examination revealed macrocephaly, severe micromelia with markedly shortened and curved femurs demonstrating a classical 'telephone receiver' appearance, a narrow thorax, and polyhydramnios. Type I thanatophoric dysplasia was suspected and subsequently supported by fetal computed tomography. Due to contextual and legal constraints regarding termination of pregnancy, expectant management was adopted. Intrauterine fetal demise occurred at 35 weeks of gestation, followed by vaginal delivery of a stillborn female neonate weighing 1940 g. Postmortem radiography confirmed the characteristic skeletal abnormalities.

This case highlights the pivotal role of prenatal imaging in diagnosing lethal skeletal dysplasias, particularly in settings where molecular testing and termination options may be limited.

Keywords: Thanatophoric dysplasia; Prenatal diagnosis; FGFR3 mutation; Lethal skeletal dysplasia; Micromelia

1. Introduction

Thanatophoric dysplasia is a rare autosomal dominant skeletal disorder with an estimated incidence ranging between 1.5 and 8 per 100,000 births. It results from activating mutations in the FGFR3 gene, leading to abnormal endochondral ossification and profound skeletal maldevelopment. The condition is considered uniformly lethal, with neonatal death occurring shortly after birth due to severe respiratory insufficiency secondary to thoracic hypoplasia.

Two clinical subtypes have been identified. Type I is characterized by severe micromelia with curved long bones and flattened vertebral bodies, whereas type II presents with relatively straight long bones and a cloverleaf skull deformity. Early prenatal recognition is essential for parental counseling and multidisciplinary management.

2. Case Presentation

A 26-year-old nulliparous woman with no significant medical or family history and no consanguinity presented at 34 weeks of gestation with symptoms of threatened preterm labor. The pregnancy had not been medically supervised

* Corresponding author: Tazi Y

before.

Obstetric ultrasound demonstrated macrocephaly with a biparietal diameter of 96 mm, severe micromelia, femoral length of 15.9 mm, humeral length of 18 mm, and markedly curved femurs producing the typical 'telephone receiver' configuration. The thoracic cavity appeared narrowed, and polyhydramnios was noted. No cloverleaf skull deformity was observed. Fetal cardiac activity was normal at the time of examination.

Fetal computed tomography confirmed macrocrania, thoracic narrowing with normal rib count, and generalized shortening and curvature of the long bones, supporting the diagnosis of type I thanatophoric dysplasia.

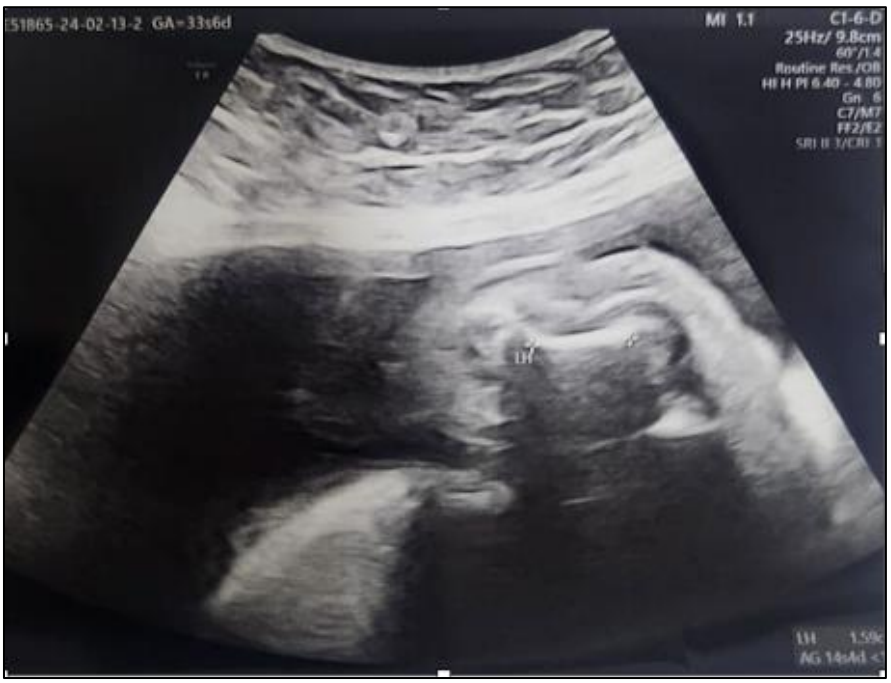

Figure 1 Ultrasound image demonstrating markedly shortened and curved long bones with a characteristic "telephone receiver" appearance, consistent with thanatophoric dysplasia

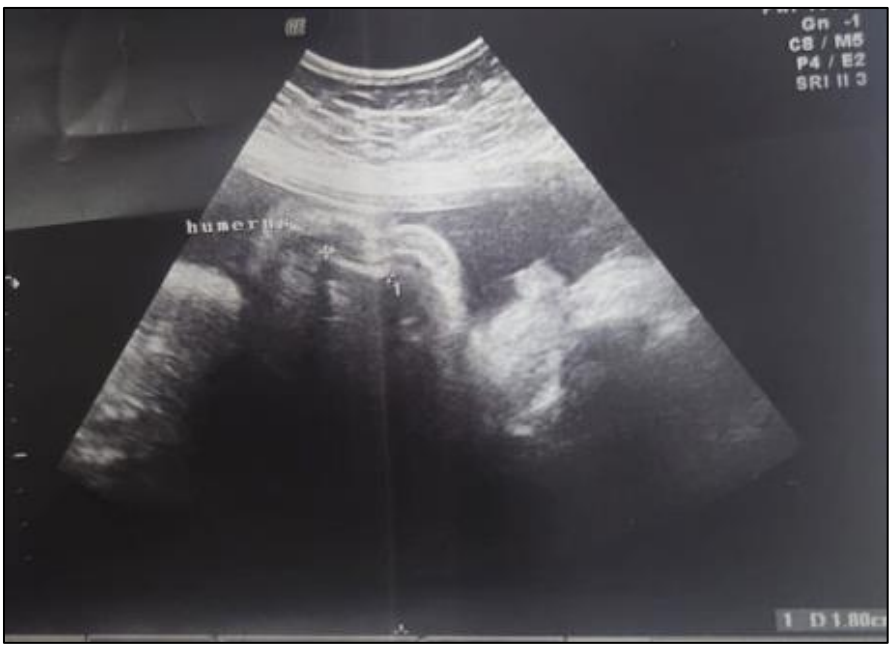

Figure 2 Ultrasound image demonstrating a markedly shortened and curved humerus in the fetus of the same patient, consistent with thanatophoric dysplasia

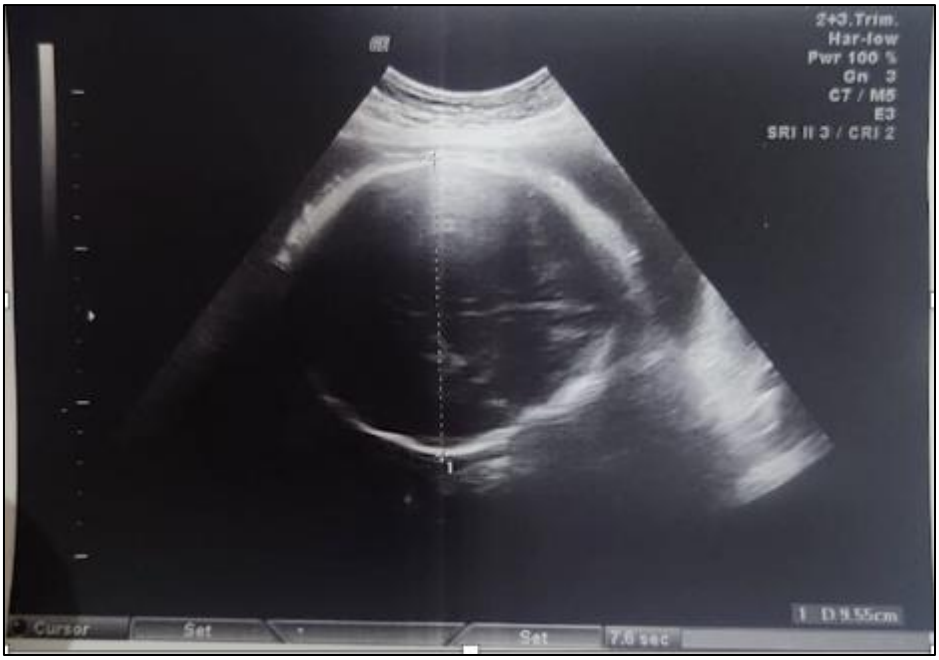

Figure 3 Ultrasound image demonstrating macrocrania with a biparietal diameter (BPD) of 95.5 mm at 34 weeks of gestation

After detailed counseling, expectant management was chosen due to local legal constraints. At 35 weeks of gestation, intrauterine fetal demise was diagnosed. Vaginal delivery resulted in a stillborn female neonate weighing 1940 g.

Figure 4 Photographs of the stillborn infant with thanatophoric dysplasia from the same patient, delivered vaginally at 35 weeks of gestation

Postmortem radiography confirmed shortened, curved femurs and humeri with flattened vertebral bodies displaying an H-shaped configuration.

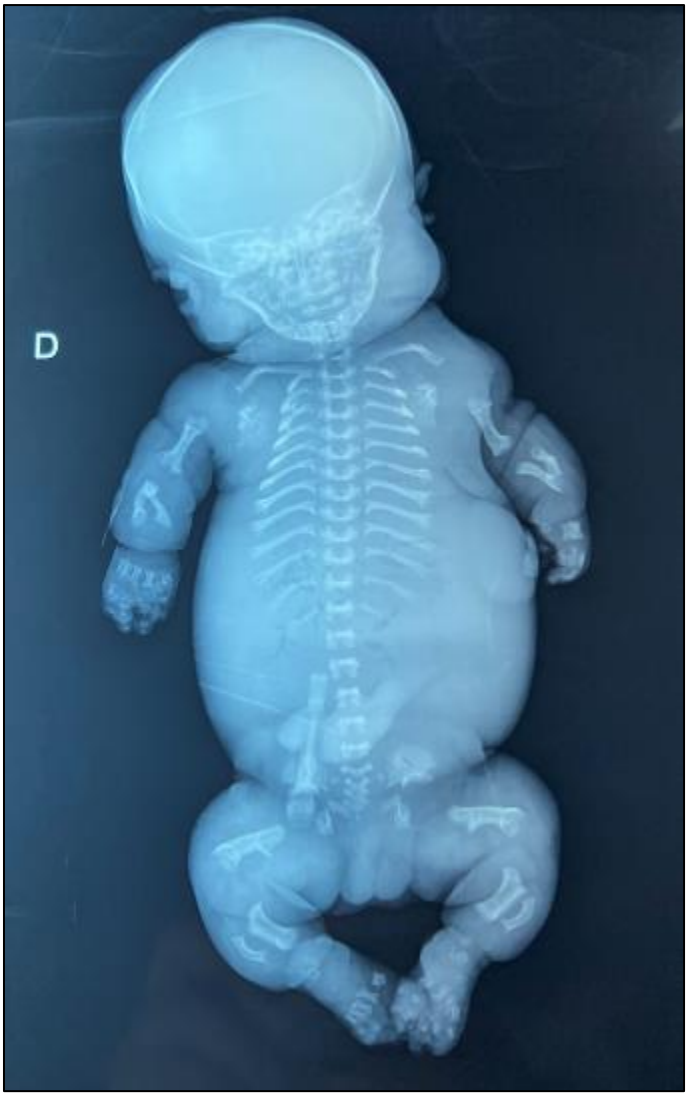

Figure 5 Postmortem radiograph of the stillborn infant with thanatophoric dysplasia from the same patient

3. Discussion

Thanatophoric dysplasia represents one of the most severe and lethal skeletal dysplasias[1].

According to Machado et al. [2], the term “thanatophoric” is derived from Greek mythology, from “Thanatos,” meaning the personification of death.

The life expectancy of newborns with NT is estimated to be about one hour after birth, according to Noe et al. Death typically results from respiratory failure caused by pulmonary hypoplasia and restricted thoracic expansion, according to Pietryga et al.[3]

The prenatal diagnosis of this malformation aims to offer a medical termination of pregnancy, as well as to prepare the couple psychologically.

Prenatal ultrasound remains the cornerstone of diagnosis, with characteristic findings including:

- Macrocephaly.
- Narrow thorax
- Extreme limb shortening.
- Hydramnios [4].

The curvature of the femurs [5] in type I provides a highly suggestive diagnostic clue.

In type II, a cloverleaf skull deformity may be observed.

Differential diagnoses include achondroplasia, osteogenesis imperfecta, achondrogenesis, and other non-lethal micromelic dysplasias.

When available, molecular confirmation through FGFR3 mutation analysis establishes definitive diagnosis. However, in resource-limited settings, imaging findings alone may guide clinical decision-making.

This case underscores the importance of systematic prenatal morphological assessment and highlights the ethical and clinical challenges encountered in environments where termination of pregnancy for lethal anomalies may not be feasible.

4. Conclusion

Type I thanatophoric dysplasia can be reliably suspected prenatally based on characteristic imaging features. Early diagnosis enables informed parental counseling and multidisciplinary planning. This report emphasizes the diagnostic value of detailed ultrasound examination and complementary imaging in the management of lethal skeletal dysplasias, particularly in low-resource settings.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

References

[1] H. Sawai et al., « National survey of prevalence and prognosis of thanatophoric dysplasia in Japan », Pediatrics International, vol. 61, no 8, p. 748-753, août 2019, doi: 10.1111/ped.13927.

[2] L. E. Machado, F. Bonilla-Musoles, et N. G. Osborne, « Thanatophoric dysplasia », Ultrasound in Obstet & Gyne, vol. 18, no 1, p. 85-86, juill. 2001, doi: 10.1046/j.1469-0705.2001.00402.x.

[3] C. Chen, S. Chern, T. Chang, C. Lin, W. Wang, et C. Tzen, « Second trimester molecular diagnosis of a stop codon FGFR3 mutation in a type I thanatophoric dysplasia fetus following abnormal ultrasound findings », Prenatal Diagnosis, vol. 22, no 8, p. 736-737, août 2002, doi: 10.1002/pd.377.

[4] N. Kouamé, A.-M. N'goan-Domoua, R.-D. N'gbesso, et A.-K. Kéita, « Diagnostic anténatal d'un cas de nanisme thanatophore type I », Imagerie de la Femme, vol. 21, no 4, p. 171-175, déc. 2011, doi: 10.1016/j.femme.2011.09.001.

[5] E. Miller, S. Blaser, P. Shannon, et E. Widjaja, « Brain and Bone Abnormalities of Thanatophoric Dwarfism », American Journal of Roentgenology, vol. 192, no 1, p. 48-51, janv. 2009, doi: 10.2214/AJR.08.1524.