

Images in Clinical Medicine

Thanatophoric Dysplasia — A Lethal Skeletal Dysplasia

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Fig 1. Antenatal ultrasonography showing narrow thoracic cage and mesocardia (altered cardiac axis)

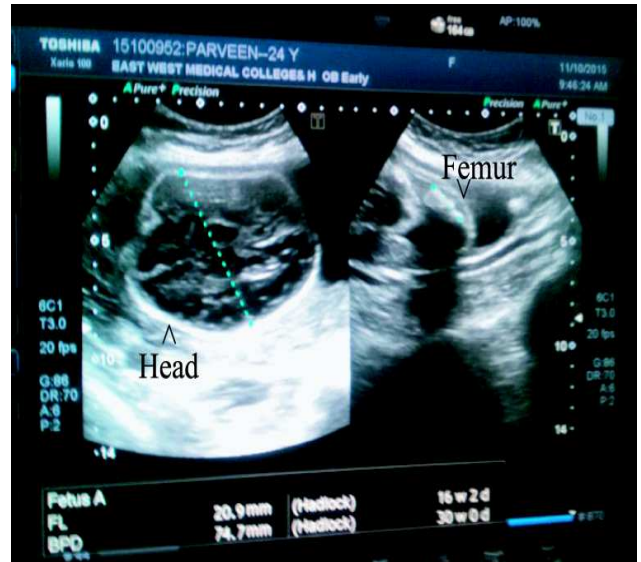


Fig 2. Antenatal ultrasonography showing macrocrania and hypoplastic fetal limbs



Fig 3. Babygram showing rhizomelic appearance with metaphyseal flaring of limbs, hypoplastic iliac bones, narrow chest with short horizontal ribs, relative macrocephaly and platyspondyly (flattening of vertebral bodies)



Fig 4. The baby just after birth

A 24-year-old pregnant woman attended the department of Radiology & Imaging in East West Medical College Hospital for ultrasonography of pregnancy profile with the history of seven months amenorrhea. Ultrasonography revealed disproportionate fetal biometry, macrocrania, hypoplastic limbs, micromelia, bowing of extremity, narrow chest, mesocardia and extra fold of skin (Fig 1, 2). These features are suggestive of thanatophoric dysplasia.

Babygram showed rhizomelic appearance with metaphyseal flaring of limbs, hypoplastic iliac bones, narrow chest with short horizontal ribs, relative macrocephaly and platyspondyly (flattening of vertebral bodies) (Fig 3).

Thanatophoric dysplasia (TD) is a lethal skeletal dysplasia. It is the 2nd most common lethal skeletal dysplasia after osteogenesis imperfecta type II.¹ The estimated incidence is at around 1:25,000–50,000.² It results from a mutation coding for the fibroblast growth receptor 3 (FGFR3) located in chromosome 4p16.3. Inheritance is thought to be sporadic.

There are two subtypes.

Type I: Marked underdevelopment of skeleton, telephone handle femurs more pronounced.

Type II: The presence of a cloverleaf skull may be a distinctive feature. This dysplasia is associated with polyhydramnios.²

Antenatal ultrasound

It may be difficult to accurately diagnose before the 3rd trimester (\approx 22 weeks).³ Prior to that it can be included in the differentials if there is a short femur length measurement.

Sonographically detectable features may include

- Relatively narrow thoracic cavity
- Short, thick, bowed tubular bones, especially lower extremity
- Thickened soft tissues of extremities
- Comparatively large head with frontal bossing
- A cloverleaf skull appearance in type II.³

Plain film

Plain radiography, if done, is usually done postmortem. Features include

- Proximal portions of the long limbs are small giving a rhizomelic appearance. Long limbs (typically

humeri and femora) have a typical "telephone handle" bowing with metaphyseal flaring.

- Iliac bones are usually hypoplastic, small squared iliac wings and may show a "trident" acetabular roof.
- Chest is narrow shaped and ribs are short and horizontal. Scapule are small.
- There are relative macrocephaly with frontal bossing, proptosis, nasal bridge flattening and Kleeblattschaedel or cloverleaf skull (with type II).²⁻⁴
- There is platyspondyly (flattening of vertebral bodies).

Prognosis

The condition is uniformly fatal within a few hours of birth either from respiratory failure or from brainstem compression from a narrow foramen magnum.⁵

Differential diagnosis

Achondroplasia

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