Prenatal Imaging of Thanatophoric Dysplasia: A Case Report

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Abstract

Thanatophoric dysplasia is the most common lethal congenital chondrodysplasia with characteristic features of narrow thorax, short rib, severe platyspondylynthia, short bowed limbs and skull deformity, etc. It is not a hereditary disorder and there is usually no family history of dysplasia. We experienced a case of thanatophoric dysplasia at 38 weeks of gestation with antenatal sonographic and abdominal radiographic findings of small thorax, short bowed extremities with surrounding thickened soft tissues and marked platyspondylynthia. Soon after delivery, the baby died and post-mortem radiographs showed the characteristic findings of thanatophoric dysplasia.

Index Words: Bones, osteochondrodysplasias 40.151, 40.152
Fetus, abnormalities 856.871, 856.873
Fetus, US studies 856.1298
Pregnancy, US studies 856.1298

Thanatophoric dysplasia is the most common neonatal short-limbed dwarfism which is incompatible with life and it has long been misinterpreted as a severe form of achondroplasia until 1967, when Maroteaux et al. (1) first described it as a definite and discrete entity.

We experienced a case of thanatophoric dysplasia at 38 weeks of gestation studied with sonography(US), maternal abdominal radiograph and post-mortem radiographs.

CASE REPORT

A 28-year-old woman visited our hospital for prenatal evaluation of her second baby. Her previous pregnancy was normal and there was no family history of dysplasia. She complained no problems except slightly weak power of fetal movement compared with her first pregnancy. Real-time US(Acuson, Mountain View, CA) with 3.5-MHz transducer showed polyhydraminos, small thorax and thickened soft tissue surrounding bowed thick long bones which are very short for the gestational age(Fig. 1a,b). Abdominal radiograph revealed severe platyspondylynthia and bowed thick long bones(Fig. 2). Soon after delivery the female baby died and post-mortem radiographs demonstrated the characteristic findings of thanatophoric dysplasia(Fig. 3).

DISCUSSION

The prevalence of thanatophoric dysplasia is one in 6,400 to 16,700(2-4) births and the inheritance pattern is sporadic. The name is de-
Fig. 1. US of fetus, gestational age of 38 weeks.
a. Small thoracic cage (small arrows) and relatively protruded-looking abdomen (large arrows) but, abdominal circumference, head circumference and biparietal diameter were appropriate for gestational age.
b. Bowed, thickened and very short femur (empty arrows) appropriate for length of 17 weeks of gestation and thickened soft tissue of extremity (black arrows).

 Derived from the Greek “thanatophoros” which means “constantly bearing death” since early death is constant and inevitable. Distinguishing US features described are polyhydraminos, narrow thorax, macrocephaly, Kleblattschädel (cloverleaf skull) deformity, short bowed thick long bones and thickened soft tissues of extremities (2-10) and the radiographic findings are characteristic for marked platyspondyly with “H” or inverted “U” shape of vertebral body, narrow thorax with short rib, macrocrania and Kleblattschädel deformity, short curved long bone, flat acetabular roof and narrow sciatic notch (1-7, 9, 10).

Our case did not show the macrocrania, hydrocephalus and Kleblattschädel deformity. Cloverleaf skull deformity is well known with thanatophoric dysplasia (2, 4, 6, 8-10) but the incidence of association is only 14% (8). This deformity consists of grotesque trilobed skull, mimicking cloverleaf appearance with prominent both temporal and vertex regions due to early intrauterine closure of coronal and lambdoid sutures, but it is not specific for thanatophoric dwarfism because homozygous achondroplasia, Apert syndrome, Pfeiffer.

Fig. 2. Plain abdominal radiograph. Marked platyspondyly (empty arrows) and thickened curved long bones of upper extremity (black arrows) and normal appearance of skull.
syndrome, Carpenter syndrome and Crouzon disease can also show this deformity(8). But with US findings of short curved limb, no family history of dysplasia associated with cloverleaf skull deformity can make the diagnosis of thanatophoric dwarfism(8).

Sonographic demonstrations of shortened and curved extremities are proofs of congenital chondrodysplasias including thanatophoric dysplasia but homozygous achondroplasia, achondrogenesis, camptomelic dysplasia, osteogenesis imperfecta(type II), asphyxiating thoracic dys trophy, hypophosphatasia and short-rib polydactyly syndrome can also show those features(4,8).

Although the reason was not certain, soft tissue thickenings in extremities were previously reported in US studies(5-7). Cremin et al. (5) proposed the hydrops accompanied with generalized circulatory failure and the disproportionate growth of soft tissue and skeleton as the etiologies.

**Fig. 3.** Post-mortem radiographs of anteroposterior(a) and lateral (b) views.
Marked platyspondyly with “H” shape of vertebral body, most characteristic to thanatophoric dysplasia and narrow thorax with short ribs, small square iliac bones, flat acetabular roofs, thickened curved very short long bones of extremities especially “telephone receiver” appearances of both femora.

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Thanatophoric Dysplasia의 산전 영상진단: 증례보고

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Thanatophoric dysplasia는 출생 후 짧은 시간 내에 사망하는 선천성 왜소성 골이형성증의 하나로서 그 빈도는 6400에서 16700명 중의 하나로 보고되고 있다. 저자들은 출산 전 임신 38주에 초음파와 단순 복부 활영으로 그 특징적 소견을 관찰하고 출산 후 곧 사망한 1예를 경험하였기에 이에 보고하는 바이다.