Case 932

Eurorad

Thanatophoric Dysplasia

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Section: Paediatric radiology
Case Type: Clinical Cases
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Clinical History:

A fetus (24-week gestation) was presented for a postmortem radiographic examination. A prenatal ultrasound (not shown) revealed a generalized micromelia, a very large skull and a small narrow chest.
Imaging Findings:

A 24-week-old fetus was presented for a postmortem radiographic examination. A prenatal ultrasound (not shown) revealed a generalized micromelia, a very large skull and a small narrow chest.
Discussion:

Thanatophoric Dysplasia (TD) is the most common presentation of lethal neonatal skeletal dysplasia. Its frequency has been described from 1.7 to 3.8 per 100.000 births. The mode of inheritance is a new autosomal dominant mutation. The genetic defect is located on chromosome 4p16. 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 a 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 height of the vertebral bodies, with sparing of the height of the pedicles. Histopathologically there is a disorganization of endochondral bone formation with lack of ordered rows of cartilage cells, a feature that is very similar to that seen in classical achondroplasia. The differential diagnosis includes Thanatophoric variants, homozygeous achondroplasia, achondrogenesis I and II, asphyxiating thoracic dysplasia and short rib-polydactyly syndromes.
Differential Diagnosis List: Thanatophoric dysplasia

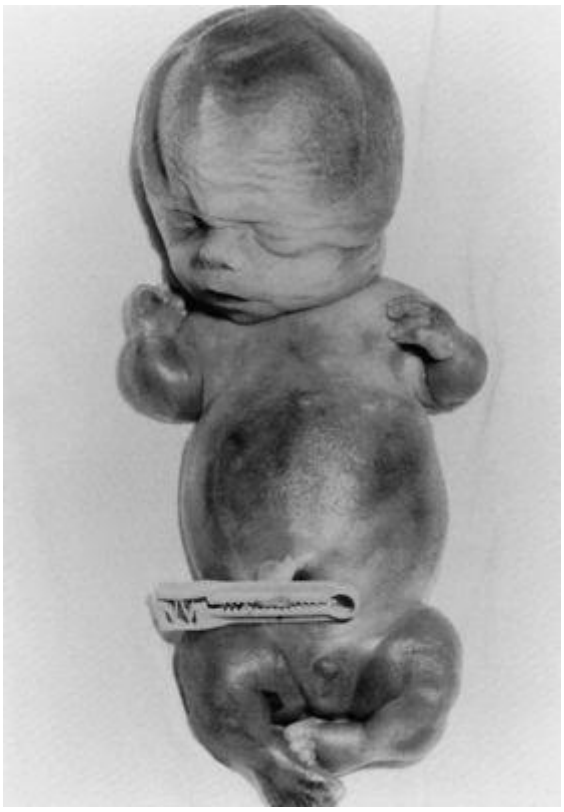
Final Diagnosis: Thanatophoric dysplasia

References:

Caffey’s pediatric X-ray Diagnosis. St Louis, Mosby Year Book, 1993; 1561-1563.
Kaufman R, Rimoin D, McAlister W et al. Thanatophoric Dwarfism. Amer J Dis Child 1970; 120: 53-57. (PMID: 4987261)
Taybi H, Lachman R., Radiology of Syndromes, Metabolic disorders and skeletal dysplasias. St Louis, Mosby Year Book, 1996; 939-941.

Figure 1

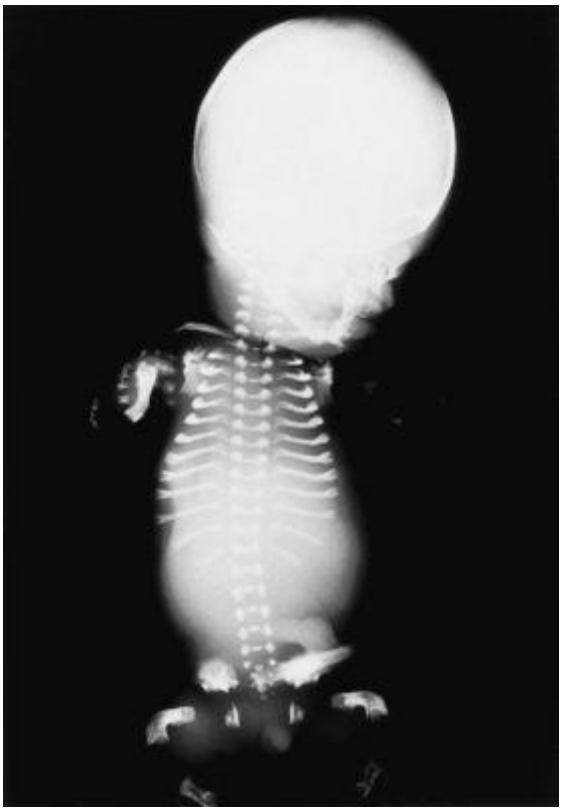
a



Description: Male fetus with disproportionate large head, small thorax and marked short-limbed dwarfism. The limbs are curved. The forehead is high with prominent frontal bossing and a depressed nasal bridge. **Origin:**

Figure 2

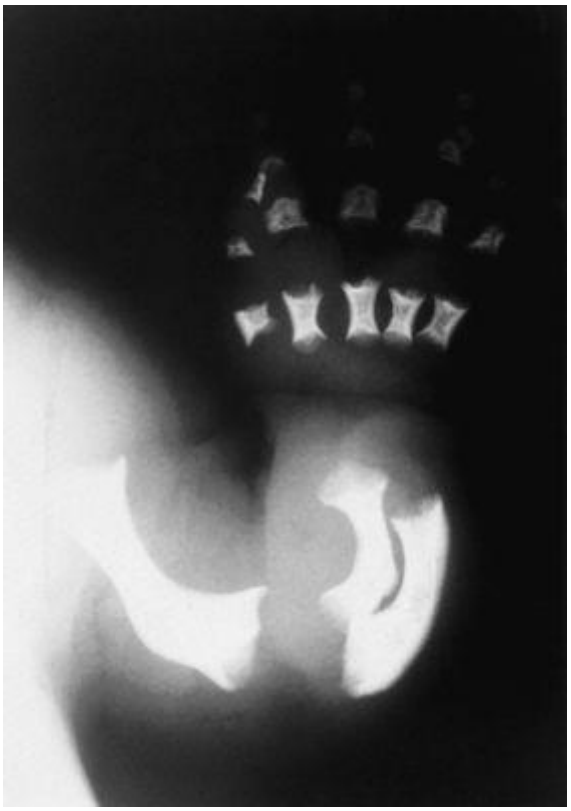
a



Description: AP view: the calvaria are relatively large compared to the short and narrow skull base and small facial bones. The chest is narrow and the ribs are very short, with cup-shaped costochondral junctions. Dysplastic appearance of both scapulae is seen. The pelvic is small with short and small iliac bones, horizontal acetabular roofs and small iliac notches. All the long bones are short with preferential involvement of the proximal segments (femora, humeri). **Origin:**

Figure 3

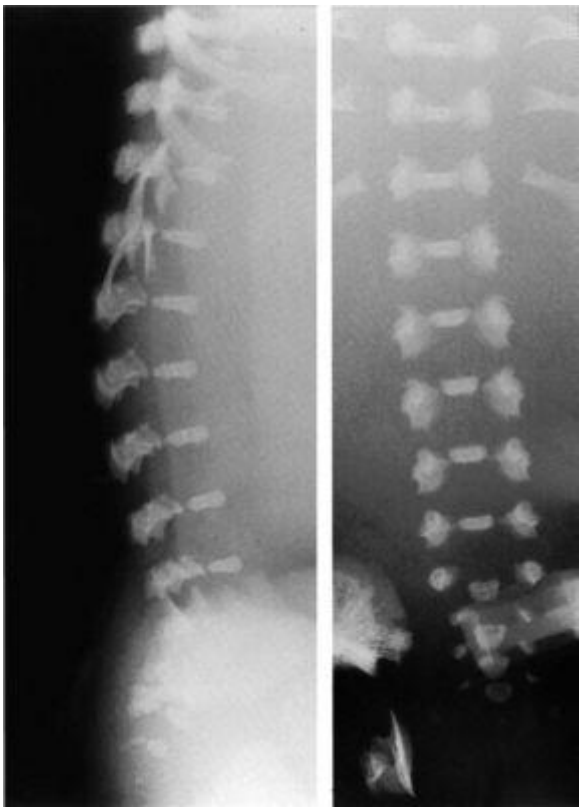
a



Description: Short and bowed long bones, having the shape of a “French telephone receiver”. Irregularity and flaring of the metaphyses are seen. **Origin:**

Figure 4

a



Description: Severe platyspondyly with apparent wide intervertebral spaces; H-shaped appearance of the vertebral bodies, and narrowing of the interpediculate distance towards the lumbosacral junction

Origin: