

DIAGNOSIS OF THANATOPHORIC DWARFISM IN UTERO

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SUMMARY

Thanatophoric dwarfism is a severe form of neonatal shortlimbed skeletal dysplasia. Most infants are stillborn or die soon after birth. This disorder has well defined radiological features which distinguish it from the other forms of neonatal dwarfism. We report two cases where short limbs were detected on sonography and a diagnosis was made on antenatal radiographs and fetography.

CASE 1

A 29-year-old multipara presented at the University Hospital, Kuala Lumpur at 26 weeks gestation. She has had two normal deliveries at this hospital, a girl in 1977 and a boy in 1978. Both children are normal and healthy. In this third pregnancy uterine size was larger than dates and hydramnios was suspected. Sonography confirmed the hydramnios and showed a single foetus with hydrocephalus and micromelia. BPD was 83 mm at 26 weeks and 114 mm at 35 weeks gestation by dates. Shortlimbed dwarfism was suspected. Antenatal radiographs and fetography were performed. These showed short curved limb bones, H-shaped vertebral bodies, large head and narrow thorax with short ribs. On the basis of these radiographic features (Fig. 1) an antenatal diagnosis of thanatophoric dwarfism was made. 250 ml of fluid was aspirated from the foetal head by percutaneous needle puncture under ultrasound

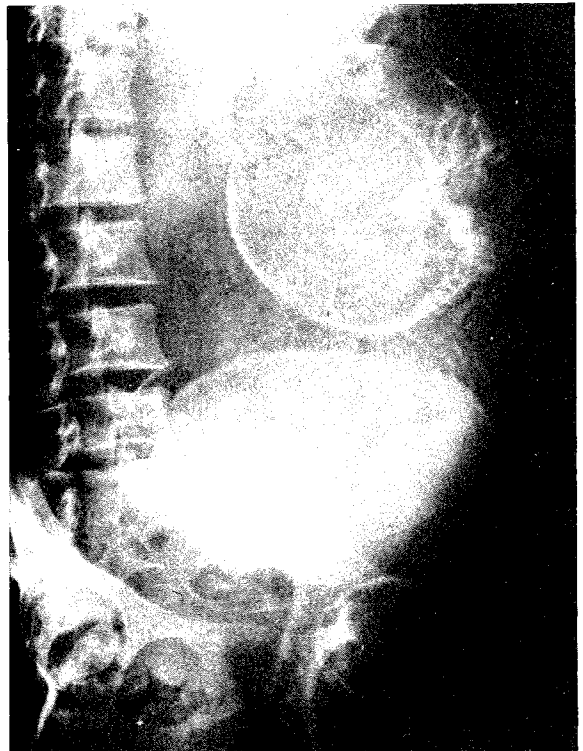


Fig. 1 Fetography of thanatophoric dwarf (Case 1) showing the short, curved limb bones, large head, narrow thorax with short ribs and H-shaped vertebral bodies.

and fluoroscopic guidance. Labour was induced the following day and an assisted breech delivery was performed. The stillborn female foetus weighed 2450 gm. The clinical features and postmortem radiographs (Figs. 2 and 3) show the characteristic appearances of thanatophoric dwarfism. Other investigations were unremarkable.

Haemoglobin was 10.4 mg%, blood group O Rh positive and VDRL test was negative. The oral glucose tolerance curve was normal. TORCHES study for maternal toxoplasmosis, rubella, cytomegalovirus and herpes CF antibodies was

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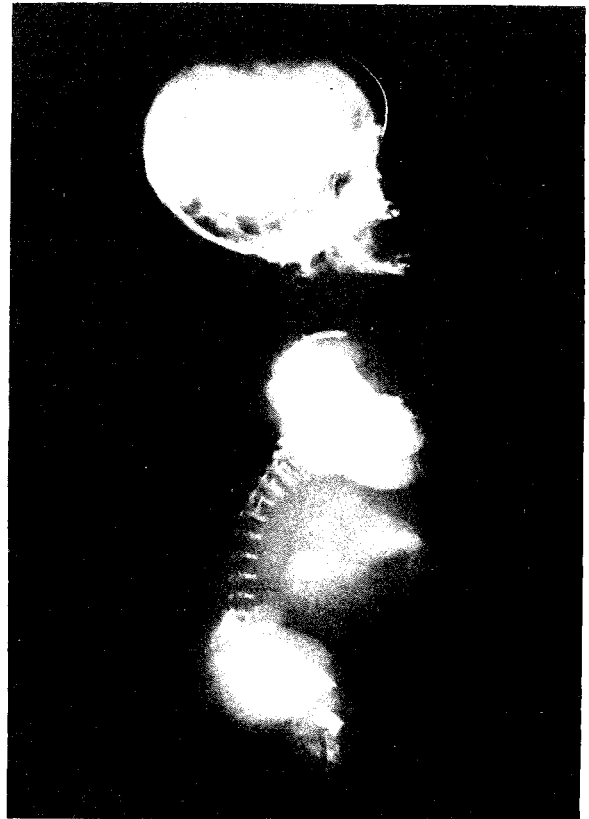
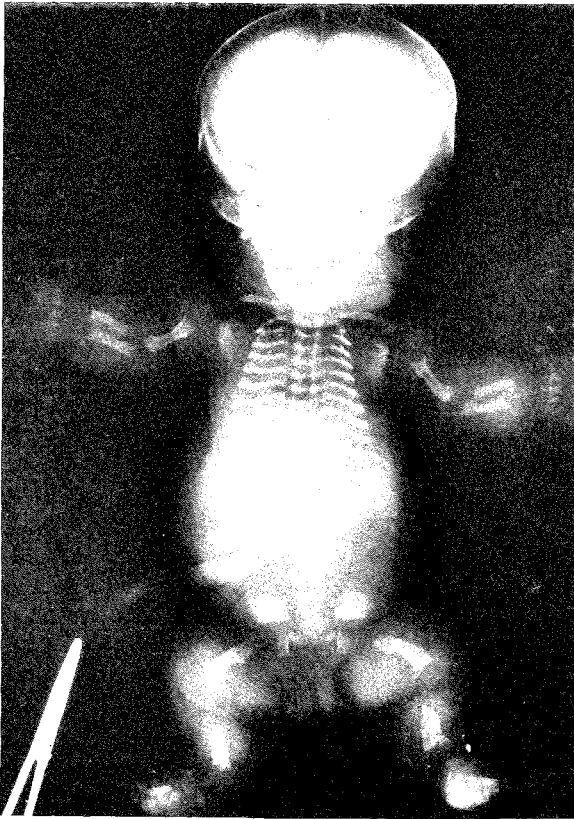


Fig. 2 & 3 Postmortem radiographs of thanatophoric dwarf (Case 1): frontal projection shows shortened and curved limb bones, telephone receiver appearance of femurs, abnormal pelvis, H-shaped vertebral bodies, narrow thorax with short ribs and large skull. Lateral projection shows thin disc like vertebral bodies with widened intervertebral spaces.

normal. The amniotic fluid for alpha fetoprotein was negative.

CASE 2

A 19-year-old primigravida was admitted to the University Hospital with right iliac fossa pain of two days duration. She was unable to give a proper menstrual history, hence the gestational period was uncertain. Uterine size was of 38 weeks gestation. Foetal parts were not easily felt and hydramnios was suspected. Sonography confirmed the hydramnios and showed a single foetus with short limbs. The presentation was cephalic and the BPD measured 85.3 mm. There was no hydrocephalus. Antenatal radiographs and fetography were performed and a diagnosis of neonatal dwarfism probably thanatophoric dysplasia was made (Fig. 4). Four weeks after admission the patient had a spontaneous vaginal delivery. The 1700 gm male baby died two hours after birth. The clinical appearance (Fig. 5) and radiographic features (Fig.

6) were those of thanatophoric dwarfism. The patient's general condition at admission was satisfactory. Blood pressure was normal, there was no pallor or oedema. Haemoglobin was 11.7 mg%. Blood group was O Rhesus positive and VDRL test was negative. Urine examination showed evidence of infection which responded well to antibiotics. Her presenting complaints of right fossa pain was most likely due to urinary infection. An oral glucose tolerance test was normal. TORCHES study for maternal toxoplasmosis, rubella, cytomegalovirus and herpes CF antibodies was normal. Serum and amniotic fluid for alpha fetoprotein were negative.

DISCUSSION

Thanatophoric dwarfism was not recognised as a distinct entity until the report of Maroteux in 1967. Since then there have been numerous reports and about 60 cases have been mentioned in the medical literature.^{1,2,3} This disorder is incompatible with life and is the most common cause of neonatal

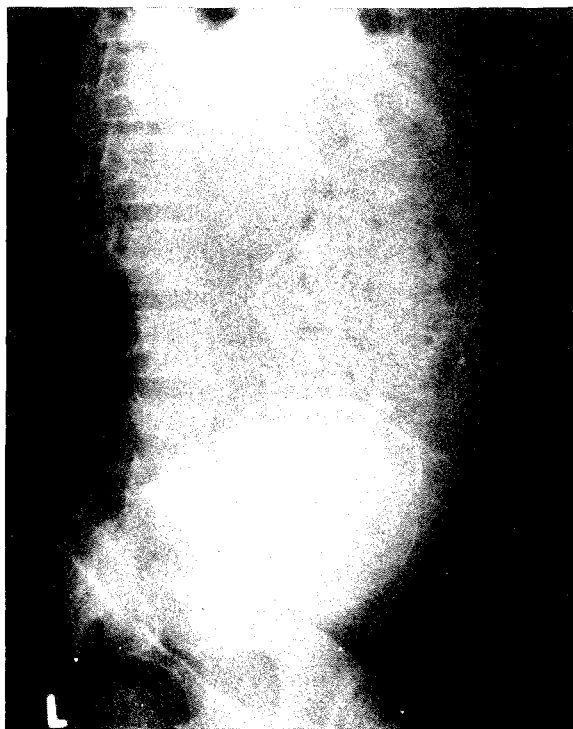


Fig. 4 Fetograph showing short limb bones, H-shaped vertebral bodies, short ribs with narrowed thorax and large head.

death among the shortlimbed dwarfs. The longest survival was of 10 weeks. ⁴ The clinical appearance of thanatophoric dwarfism is quite characteristic. The limbs are very short and pug-like. The trunk is of normal length but the thorax is markedly narrowed and the abdomen is protuberant. The arms are outstretched, the legs are externally rotated and the thighs are abducted. There is generalised soft tissue thickening and the skin is oedematous. The head is usually disproportionately large (hydrocephalus). There is depression of the nasal bridge with frontal bossing. Head measurements may be normal as was in one of our cases. On the other hand, clover-leaf type of skull deformity have been reported. ⁵

The radiological features of thanatophoric dwarfism are distinctive. A striking feature is the discrepancy between the relatively long, narrow trunk and the markedly short limbs. The cranial vault is usually well mineralised but there is prominence of the forehead and depression of the nasal bridge. The facial bones are small in comparison to size of calvarium. Hydrocephalus is usually present, but both normal size skull and



Fig. 5 Clinical appearances of thanatophoric dwarf (Case 2). Note the large head, short narrow thorax, protuberant abdomen, markedly short limbs and generalised oedema.

clover-leaf deformity have been reported. The appearance of the vertebral column and limbs are quite characteristic. A lateral view of the spine shows markedly decreased height of the vertebral bodies (thin disc like) with widened intervertebral spaces. On the frontal projection the vertebral bodies appear H-shaped. The long bones of the limbs are very short, broad and curved. There is flaring of the metaphyses and show irregular thornlike projections. The femur has a distinctive telephone receiver appearance. The short tubular bones are also decreased in length and show slight metaphyseal irregularity. The pelvis is small with markedly short iliac and ischial bones. The thorax is narrow in both the antero-posterior and transverse diameters owing to the short ribs. The rib ends are wide and cupped. The scapulae are small and the clavicles are normal.

The radiological features of thanatophoric dysplasia are diagnostic and there is usually no difficulty in differentiating it from the other forms of neonatal dwarfism. Radiographs of homozygous

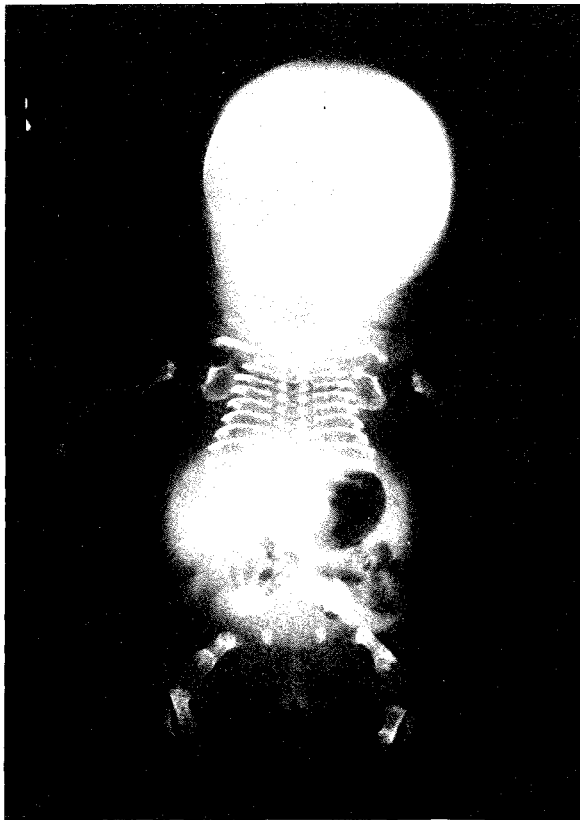


Fig. 6 Postmortem radiograph of Case 2 showing narrow thorax with short ribs, short curved limb bones, abnormal pelvis and H-shaped vertebral bodies.

achondroplasia may show some resemblance but the appearance of the spine and femur in thanatophoric dysplasia is distinctive.² In achondrogenesis there is severe unossification of the lumbar spine, sacrum, pubic and ischium.^{6,7}

With the introduction of sonography and of serum and amniotic fluid alpha fetoprotein determination it is now possible to suspect certain foetal defects at an early gestational age. In selected cases midtrimester amniography has been advocated by Balsam and Weiss.⁸ These examinations were done at 15-20 weeks after the last menstrual period in patients with elevated amniotic fluid alpha fetoprotein and where abnormality or suspected abnormality was detected on sonography. We prefer fetography as the oily contrast medium used gives a satisfactory coating and the external topographic appearances are clearly delineated. Foetal abnormalities such as deformed limbs or meningocele mass can be clearly demonstrated by this method.

Antenatal diagnosis of thanatophoric dysplasia was first reported by Campbell.⁹ O'Malley *et al*¹⁰ reported the first case detected by ultrasound which was confirmed by antenatal radiographs. Further reports of sonographic diagnosis of thanatophoric dwarfism were by other workers.^{5,11,12} Ultrasound findings of short limbs with large head particularly in cases of hydramnios should suggest neonatal shortlimbed dwarfism. Precise measurements of foetal femoral length by sonography has been advocated by Filley *et al*¹³ as a fairly accurate method for early detection of shortlimbed dwarfism. Severe foetal abnormalities represent an indication for midtrimester termination. Their early detection will allow early induction of labour.

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