

## The Thanatophoric Dwarf

### A Report on Two Cases

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**Key Words.** Thanatophoric dwarfism · Lethal form of nanism · Radioulnar synostosis · Cloverleaf skull · Craniosynostosis

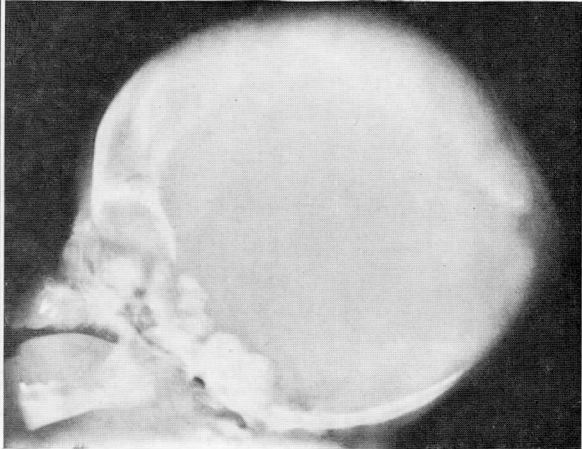
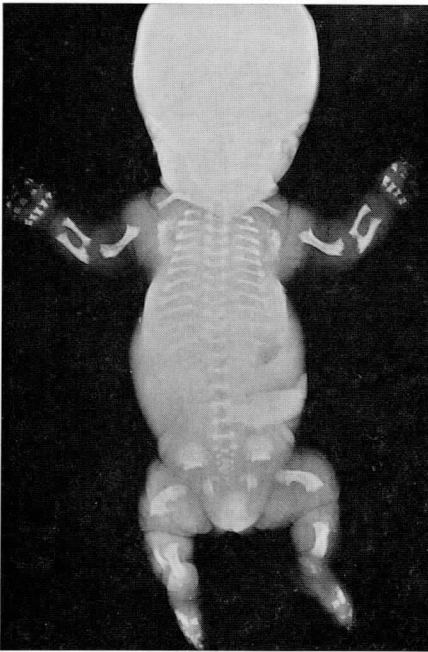
**Abstract.** Two cases of thanatophoric dwarfism are presented. At external physical examination this anomaly is not readily distinguishable from achondroplasia and other forms of congenital disproportionate dwarfism. Radiologically, however, characteristic skeletal changes are demonstrable. The diagnosis can be made *in utero*. The clinical course is invariably fatal. The hereditary aspects, of importance for genetic counseling of parents, are still unexplained.

The patients discussed show the typical thanatophoric skeletal changes in combination with a form of synostosis, i.e. bilateral radioulnar synostosis in one case, and synostosis of the coronal sutures and the lambdoid suture in the other. Emphasis is placed on the importance of radiological examination in cases of unexplained hydramnion and perinatal death.

### Introduction

In the classification of the European Society of Pediatric Radiology, the thanatophoric dwarf (from the Greek θάνατος = death, φέρω = to carry) is brought under the heading of constitutional osteochondrodysplasias of unknown pathogenesis, manifest at birth [2]. Achondroplasia is the classical representative of this group, with which the other types show more or less close similarity in appearance. Only in the past few decades have some of these types been differentiated from achondroplasia and recognized as separate entities, mainly as a result of progress made in paediatric radiology.

The thanatophoric dwarf was first described by MAROTEAUX and LAMY in 1967, and over 50 cases have since been published. The external features of 'severe' achondroplasia are visible at birth. The head is relatively large; the



*Fig. 1.* Patient 1. Plain X-ray. Conspicuous disproportionate dwarfism. Very short metacarpals. Radioulnar synostoses clearly visible.

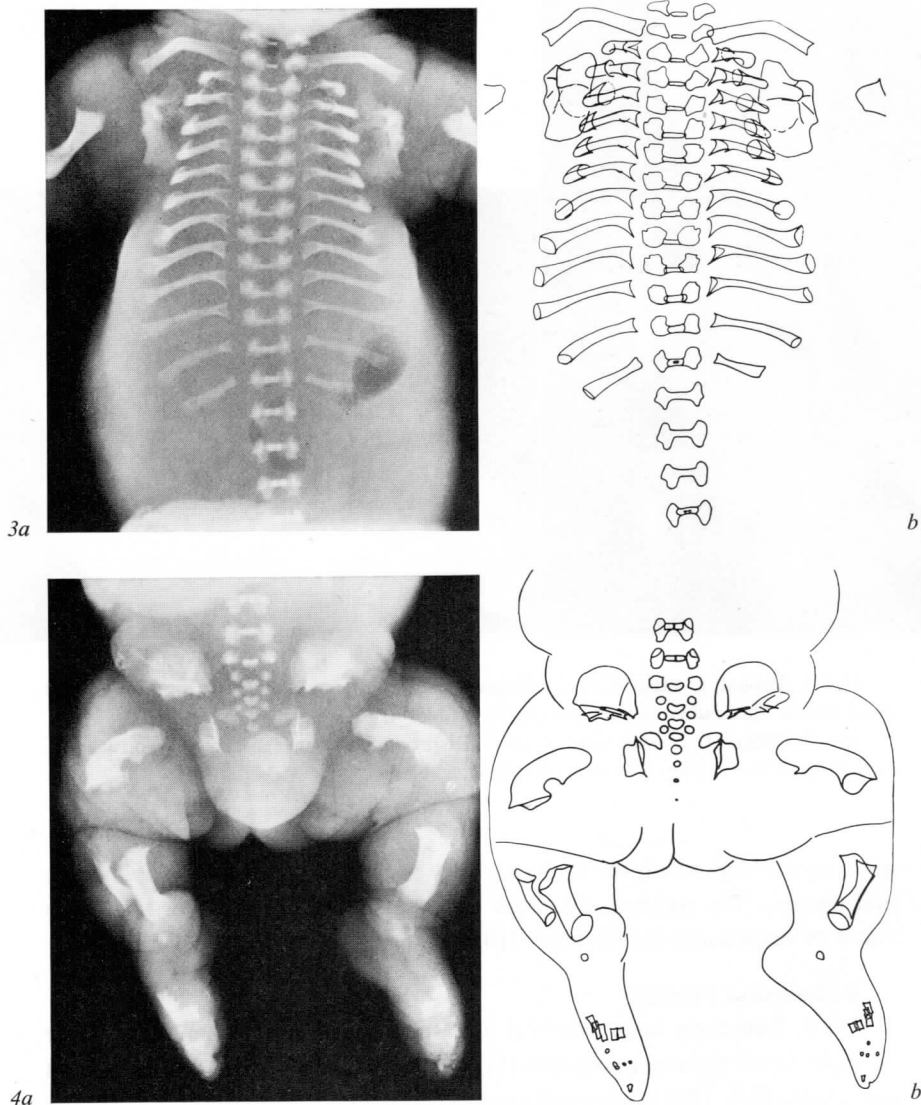
*Fig. 2.* Patient 1. Lateral view of skull. Disproportion between cerebral and visceral cranium and short, steep base of skull clearly visible.

nasal root is depressed; the limbs are very short; the thorax is relatively long and narrow. The patients die either immediately or shortly after birth as a result of respiratory insufficiency [10].

#### *Radiological Findings*

*Skull.* Relatively large cerebral and small visceral cranium. Short base of skull. Small foramen magnum [11]. A so-called cloverleaf skull is seen in some cases [13]. This is a cranial anomaly based on synostosis of the coronal sutures and the lambdoid suture with, as characteristic features, closely spaced petrosal bones and laterocaudal protrusion of the mesocranial fossae. This anomaly can be present in a more or less pronounced degree.

*Thorax.* Narrow, slender thorax. Short ribs widening towards the costochondral junction. The scapular bones are virtually square, with an irregular inferior contour.



*Fig. 3. a* Patient 1. Chest X-ray anteroposterior view. Short ribs, narrow chest. Some air visible in bronchi, stomach and duodenum. *b* Schematic drawing of patient 1. Not all the normal nutrient foramina in the vertebral bodies have been drawn.

*Fig. 4. a* Patient 1. Pelvis and lower limbs. Square iliac bones with very narrow sciatic notch. Short bowed femora, tibiae and fibulae with thorn-like processes. Short metatarsals. *b* Schematic drawing of patient 1, also indicating the demarcation of the markedly plicated soft tissues.

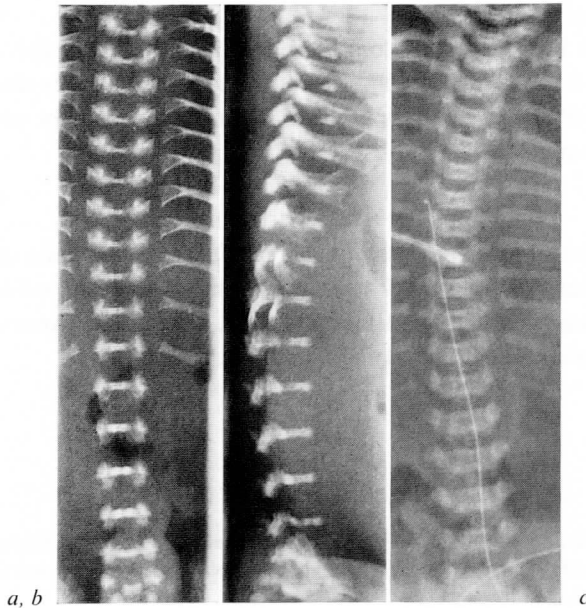


Fig. 5. *a* Patient 1. Vertebral column, anteroposterior view. U-shaped and H-shaped vertebrae with very flat ossification centre. Note broad medial costal ends. *b* Lateral view. Extreme disproportion between height of vertebral centres and that of interspaces is evident. *c* For comparison, a normal vertebral column after a pregnancy of about the same duration.

*Vertebral column.* The ossification centres of the vertebral bodies are strikingly flat, with large interspaces. The centres of the dorsal vertebral segments are of normal height. This combination produces the typical U-shape and H-shape of the vertebrae in the anteroposterior view.

*Pelvis.* The alae of the iliac bones are square; the acetabular roofs are flat, with irregular spurring; the sciatic notches are narrow; the sciatic bones show pointed ends.

*Limbs.* The long bones are bowed, broad and very short; the metaphyseal ends are flaring and sometimes show thorn-like projections; the metacarpals, metatarsals and phalanges are very short and broad.

The radiological findings are so unmistakable that the condition can be diagnosed *in utero* [1].

### *Pathological Anatomy*

There have been few pathological anatomical studies of this condition. According to RIMOIN *et al.* [8, 9], the chondral architecture is disorganized.

The achondroplasia involves an ossification disorder of the primordially normal cartilage.

### *Heredity*

The disorder in skeletal development results from a genetic defect. This can be hereditary or a result of spontaneous mutation. The mode of transmission of thanatophoric dwarfism is obscure, although the transmission of several even more exceptional dysplasias is known [12].

### *Case Reports*

*Patient 1.* R.V., born 5th March 1973 in the 32nd week of pregnancy. Hydramnion may have existed. Mother gravida II, para II, abortion I. No information available on family history and consanguinity, if any.

At birth the Apgar score was 2; weight 1,160 g; height 32 cm. There was marked asphyxia. There were visible congenital anomalies consistent with achondroplasia. The infant died shortly after birth.

The characteristic radiological changes of thanatophoric dwarfism were found (fig. 1-5;) in addition there was bilateral radioulnar synostosis (fig. 1). The postmortem revealed the external characteristics of achondroplasia. The lungs were hypoplastic and not expanded, the internal organs were otherwise normal. Small foramen magnum and flat pelvis. Microscopic examination of vertebral column and femur revealed changes similar to those described under the heading 'severe, fatal form of achondroplasia' [7] (fig. 6).

*Patient 2.* D.D., born 30st June 1974 in the 35th week of pregnancy. There was hydramnion. Mother gravida II, para I. No available information on family history and consanguinity, if any.

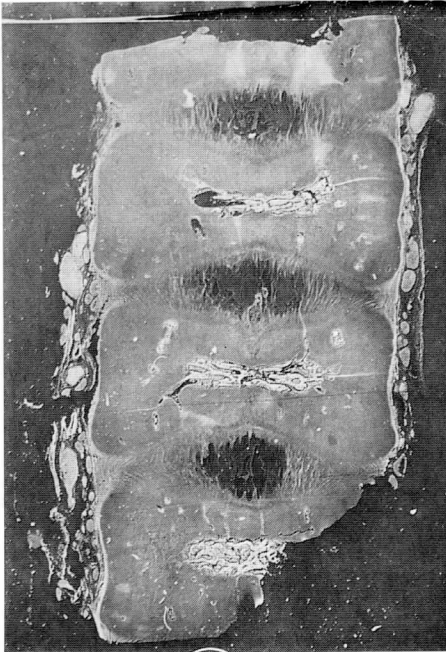
At birth the Apgar score was 8, weight 2,060 g; height 35 cm. The infant presented the appearance of an achondroplast. The initially adequate respiration deteriorated; the features

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*Fig. 6. a* Patient 1, microscopic specimen of a few thoracic vertebrae in frontal section. Flat ossification centres, the vertebrae consisting largely of cartilage. In between, disc material. *b* Patient 1, right femur. Irregular trabecular structure. Large cartilaginous capsules at ends.

*Fig. 7.* Patient 2. Plain X-ray. Note striking resemblance to patient 1. More air in intestine because the infant lived longer.

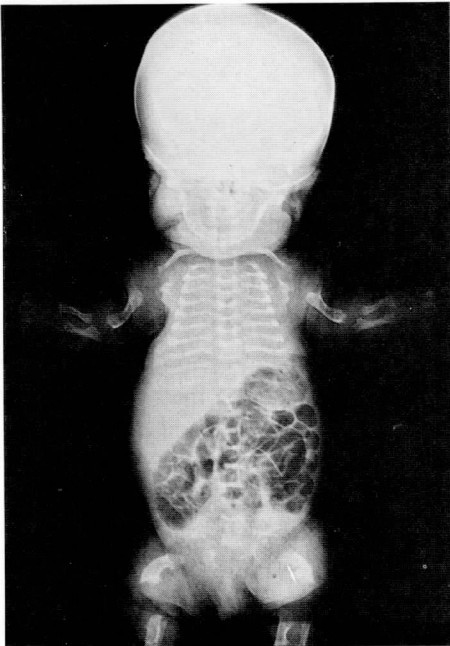
*Fig. 8.* Patient 2. Skull, anteroposterior view. Petrosal bones are close together. Slight lateral protrusion of temporal bone with, above it, a small indentation.



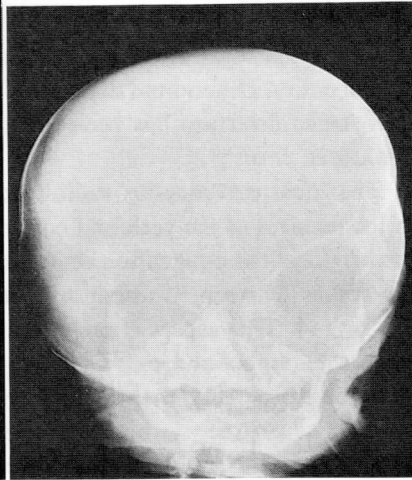
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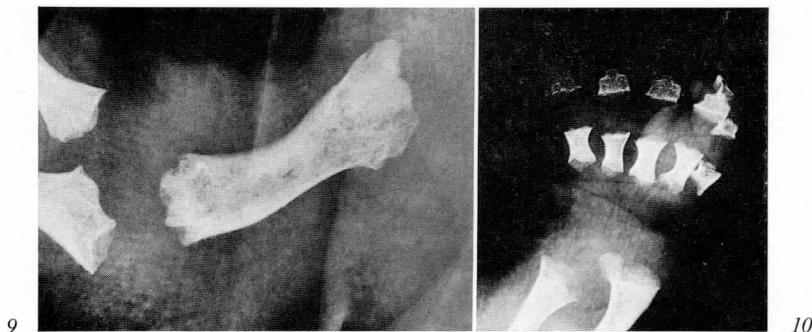


Fig. 9. Patient 2. Humerus, detail. Short thick bone with irregular trabecular structure.

Fig. 10. Patient 2. Hand, detail. Short broad metacarpals. The width of the basic phalanges exceeds their length.

of hyaline membrane disease developed, with respiratory insufficiency in which the narrow chest also played a role. The infant died on the 4th day after birth.

Radiological examination revealed skeletal changes characteristic of thanatophoric dwarfism (fig. 7, 9, 10), with in addition a mild degree of cloverleaf skull (fig. 8). The post-mortem revealed external signs of achondroplasia. Arm length 8 cm; leg length 10 cm. Hyaline membranes were found in the hyperaemic lungs. The internal organs were normal apart from hepatosplenomegaly.

### Discussion

Thanatophoric dwarfism is now generally regarded as a clinical and radiological entity. Differential diagnosis from achondroplasia and other forms of micromelic dwarfism has been discussed in detail by LANGER *et al.* [5] and SPRANGER *et al.* [12].

The most conspicuous radiological difference from achondroplasia lies in the features of the vertebral column. In achondroplasia the ratio between the height of the ossification centres of the vertebral bodies and the height of the spaces between, is about 1:1; in the thanatophoric dwarf this ratio is 1:3 or 1:4. This aspect is often erroneously referred to as platyspondylisis (cf. fig. 6). In achondroplasia, moreover, the long bones are less extremely short and bowed, and the metaphyseal ends do not show thorn-like processes. The length: width ratio of the metacarpals, metatarsals and proximal phalanges is clearly positive. The chest is usually of normal shape. Radio-

logically, the homozygotic form of (dominantly inherited) achondroplasia has a more or less intermediate position but need not pose any problems if we bear in mind that both parents should be achondroplastic dwarfs.

A cloverleaf skull can occur, not only in association with thanatophoric dwarfism but also as an isolated phenomenon [3]. So far as we know, radio-ular synostosis has been observed only once in a thanatophoric dwarf [4].

One may be tempted to philosophize about a common basis of synostoses in connection with thanatophoric dwarfism, but for the time being there are no factual arguments. It is evident, however, that precisely in the case of rare anomalies an optimally exact diagnosis is a prerequisite for prognosis and for genetic counseling. Radiology is an indispensable aid in this respect.

Finally, it may be pointed out that radiological examination can reveal unsuspected changes not only in osteogenic dysplasias but also in cases of unexplained hydramnion and perinatal death of obscure origin. This is illustrated by the above case reports.

### Acknowledgement

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