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Thanatophoric dwarfism in a Nigerian child

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Summary

We describe a lethal variety of congenital short-limbed dwarfism (Thanatophoric dwarfism) characterized by marked shortening of the extremities, macrocephaly with associated hydrocephalus and a narrow thorax, delivered by a 25-year-old woman in our hospital. Experience with this form of abnormality is rare and even an isolated case report will heighten awareness and assist with further management and counselling of parents.

Résumé

On décrit une variété congénitale mortelle de nanisme avec raccourcissement des membres (la nanisme thanatephore) caractérisé par un raccourcissement marqué des extrémités, une macrocéphalie associée à une hydrocéphalie, un thorax étroit, observée chez un nouveau né mis au monde par une femme de 25 ans à notre hôpital. Les cas de cette espèce sont rares et le rapport de ce cas isolé mettra en lumière certains points et aidera à traiter les cas ultérieurs et à conseiller les parents.

Introduction

Thanatophoric dwarfism (TD), a lethal variety of congenital short-limbed dwarfism derives its name from the Greek *Thanatos*, death; and *Phoros*, bearing, (Maroteaux, Lamy & Robert, 1967). The disorder is characterized by marked shortening of the extremities, a relatively large head with associated hydrocephalus and a narrow thorax. Respiratory distress and asphyxia secondary to chest deformity and pulmonary hypoplasia occur immediately following delivery and death is usual a few hours after birth.

The aetiology is largely unknown but a Mendelian autosomal recessive inheritance is thought likely following the birth of affected siblings to normal parents (Thomson, Reynolds & Cruickshank, 1982).

We recently encountered a case of TD which we consider worth reporting because of its rarity and because, to our knowledge, no such case has ever been reported in an African child.

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Report of case

A 25-year-old woman with two previously normal pregnancies registered to have her third pregnancy supervised in the University of Ilorin Teaching Hospital at about 20 weeks' gestation. Initial clinical evaluation together with routine laboratory investigations including VDRL test revealed no abnormalities. Polyhydramnios became evident at 30 weeks' gestation and this necessitated a request for an abdominal radiograph. This revealed a single foetus with some major malformations. The limbs were very short and the femur hook-shaped (Fig. 1), suggesting TD.

After detailed explanation of the condition, the patient consented to termination of her pregnancy. Labour was induced by artificial rupture of the membranes and pitocin stimulation. A live but grossly deformed male infant weighing 1.9 kg was delivered (Fig. 2). He had deformed short limbs, narrow chest, enlarged head and low-set ears. The breathing was shallow. He was admitted into the special care baby unit where he died a few hours later having failed to sustain a good respiratory effort.

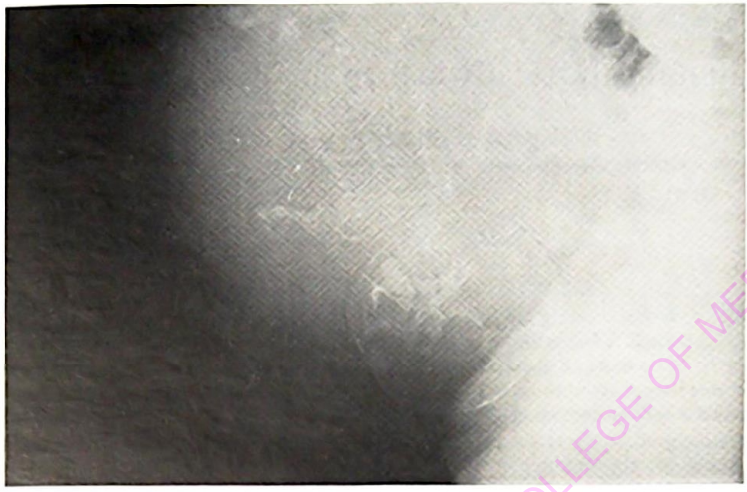

Fig. 1. Lateral radiograph of pregnant abdomen. There is a single foetus presenting cephalic, with multiple deformities of the long bones. Note ground-glass haziness from hydramnios.

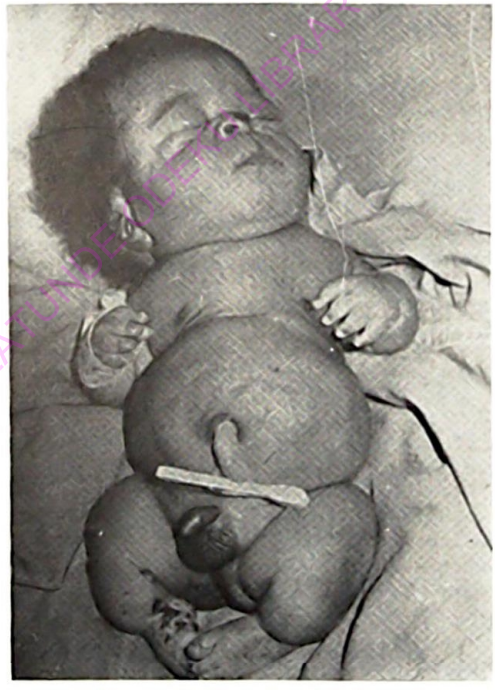

Fig. 2. Clinical photograph showing short deformed limbs, narrow chest, low-set deformed pinna, and slight macrocephaly.

Post-mortem babygram showed flaring of the metaphyses of the long bones and hook-like deformity of the femora (Fig. 3). The vertebral bodies were markedly flattened with the disc spaces unduly prominent (Fig. 4). Autopsy revealed additionally hypoplastic lungs but normal heart and abdominal viscera.

There was nothing contributory in the family review. The father was aged 34 years at the time of the child's conception. The marriage was not consanguineous. No other members of the family presented with similar abnormality.

Discussion

Thomson *et al.* (1982) gave a summary of the major clinical and radiological features of skeletal dysplasias. The present case satisfied all the cardinal features of thanatophoric dwarfism, namely, marked shortening of the extremities, characteristic facie with relatively large head, depressed nasal bridge and narrow thorax.

The radiographs also showed features already documented by Moore and Banik (1980); these include generalized micromelia, severe bowing of the femur giving the telephone receiver appearance, flaring of the metaphyseal ends of the long bones, narrow chest with short ribs and platyspondyly in which the vertebral bodies become flattened with relative increase in disc space height.

Some other less specific skeletal anomalies like elongated transverse diameter of the pelvis with horizontal acetabular roof and clover leaf skull have been described in TD (Moore & Banik, 1980).

Prognosis for survival is very poor. The

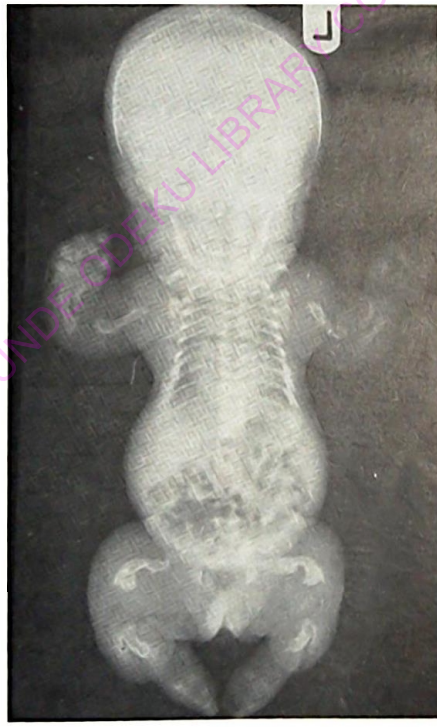

Fig. 3. Post-mortem antero-posterior radiograph. Note deformity of the long bones, particularly severe bowing of femur, resembling telephone receivers.

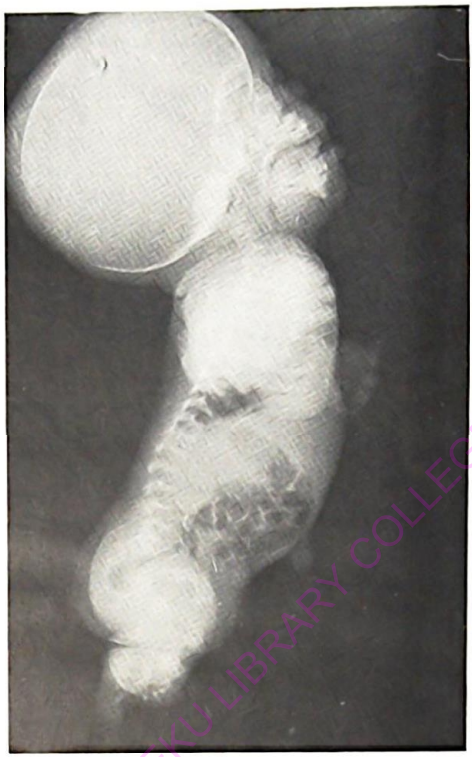

Fig. 4. Post-mortem lateral radiograph. There is severe platyspondyly and relative increase in disc space width.

longest reported case lived for only 10 weeks (Moir & Kozlowski, 1976). Cardio-respiratory insufficiency accounting for early neonatal deaths in TD was responsible for the death of our patient. In spite of its rarity, thanatophoric dwarfism has such distinctive features that it is unlikely to be confused with the other common forms of skeletal dysplasias like achondroplasia and osteogenesis imperfecta which are compatible with survival beyond the neonatal period (Harris & Patton, 1971).

Acknowledgment

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