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Thanatophoric Dwarfism

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Until recently thanatophoric dwarfism, a short-limbed dwarf condition, was misinterpreted as severe immature form of achondroplastic dwarfism. In 1967, Maroteux *et al.*(1) stated that thanatophoric dwarfism was discrete entity; the condition is rare with a prevalence of 1 in 6400 population.

Case Report

A stillborn baby was delivered to a primigravida at 40 weeks gestation. During the antenatal period the height of uterus was less than period of gestation and the presentation was breech. *In utero*, ultrasonography showed a single live fetus with biparietal diameter of 92 mm, hyperextended spine with platyspondyly, narrow thorax with pulmonary hypoplasia, normal abdominal contents, posterior fundal position of placenta, very small upper and lower extremities with bowing of long bones and excessive liquor. A diagnosis of thanatophoric dwarfism and asphyxiating thoracic syndrome was considered.

There was no history of abortion, dwarfism in parents or in other family members, consanguinity or exposure to potential teratogenic agents. Mother's 'TORCH' test was positive for Rubella and CMV antibodies. Radiographic examination of the stillborn showed disproportionately short extremities in relation to trunk. The skull was large showing frontal bossing, spine showed platyspondyly with H-shaped vertebra, the thorax was triangular in shape, tubular bones were short with bowing of long bones and slight metaphyseal irregularity and flaring.

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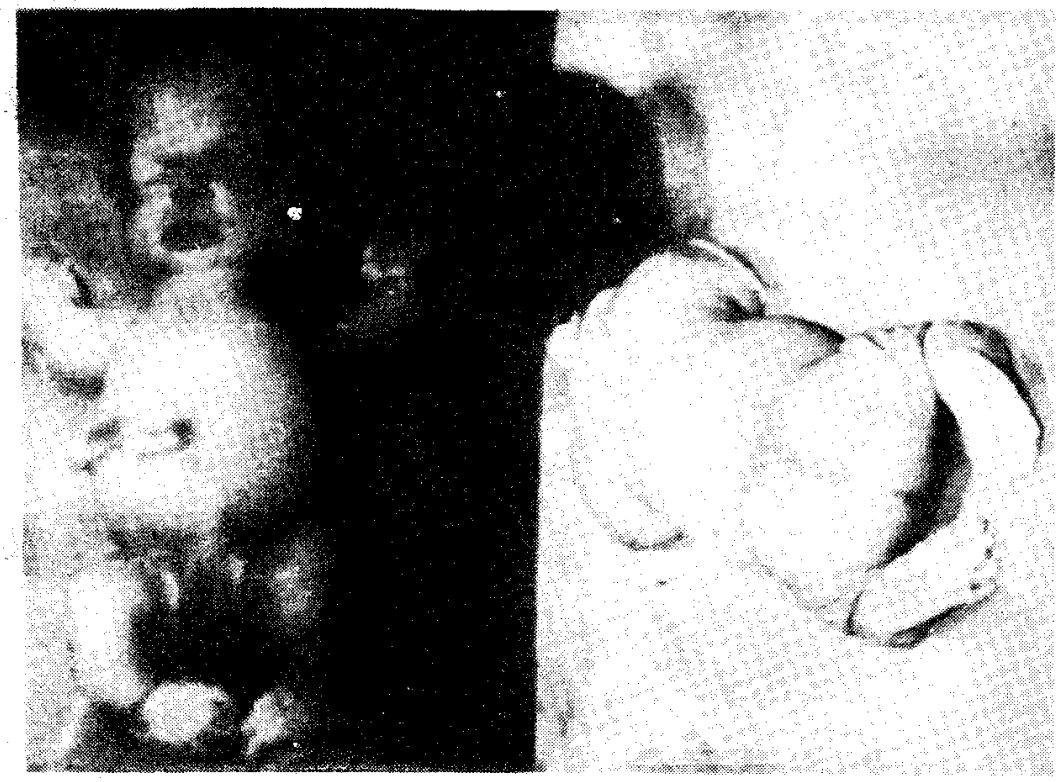

Fig. 1. Thanatophoric dwarf showing deep skin folds, depressed nasal bridge and bell shaped thorax.

Autopsy Finding: The baby weighed 2 kg, length was 32 cm and head circumference 34.5 cm with frontal bossing. The nasal bridge was depressed. The extremities were short, the upper extremity measured 9.5 cm (normal–17.8 cm), arm length 4.0 cm (normal–7.0 cm) and forearm length being 3.0 cm (normal–5.8 cm)(2). The lower extremities measured 11.5 cm (normal–18.9), thigh length 4.5 cm (normal–8.4 cm), leg 5.5 cm (normal–6.6 cm)(2). The fingers were small and thick with deep skin folds associated with increased subcutaneous fat (*Fig. 1*). The thorax was bell-shaped (narrow in the upper part and broadened near the abdomen). The chest circumference was 20 cm.

The lungs were unexpanded showing congenital pulmonary atelectasis. The lungs, heart, thymus, liver, pancreas, kidneys and adrenals were smaller than normal for the corresponding weight of the baby.

Humerus revealed wide cartilagenous cap with haphazard arrangement of chondrocytes throughout the width of cartilage. Cord like arrangement of chondrocytes was absent even adjacent to growth zone (*Fig. 2*). Provisional calficiation spicules were irregularly distributed along the growth zone.

Vertebral bodies were flattened. Cut surface of the body was completely cartilagenous grossly which was confirmed on microscopic examination.

The ribs were short and grossly showed bead-like thickening anteriorly. The cartilagenous portion of the rib was larger with costochondral junction lying towards ante-

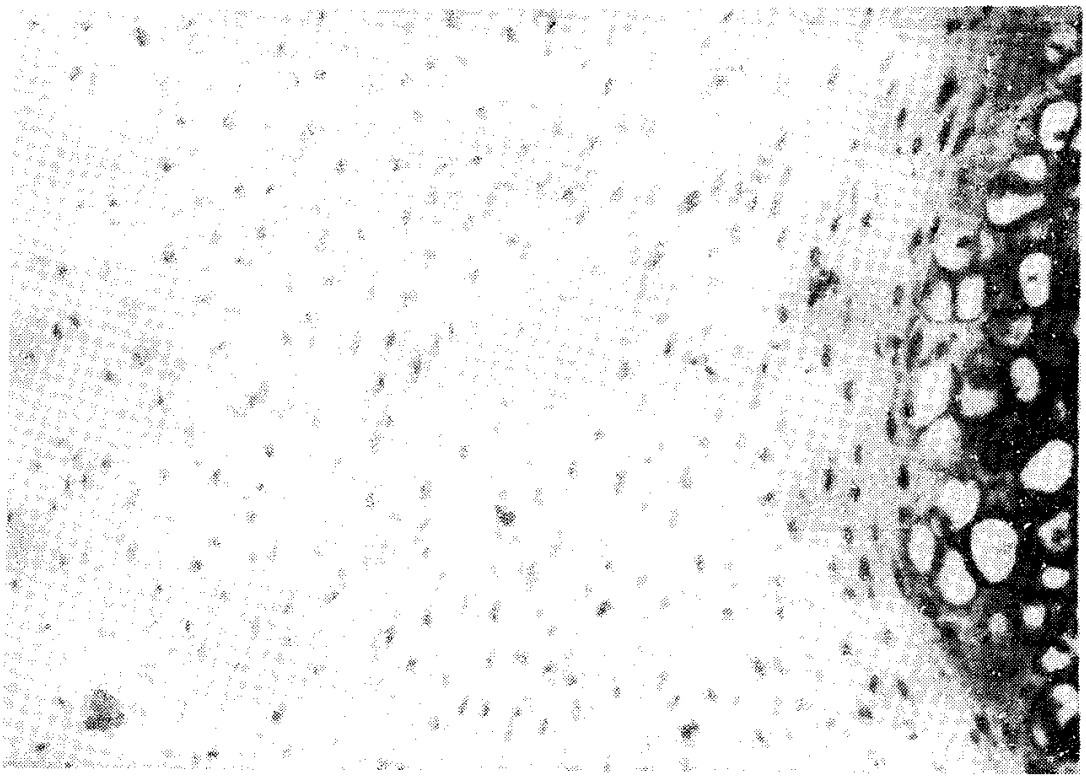

Fig. 2. Haphazard arrangement of chondrocytes (H&E, 60X).

rior axillary line. Microscopically rib also revealed haphazard arrangement of chondrocytes. A final diagnosis of thanatophoric dwarfism was made.

Discussion

There are many causes of short limb dwarfism manifesting at birth which can be distinguished from each other by their clinical and radiographic features(3-5). However, achondrogenesis, achondroplasia and asphyxiating thoracic dystrophy pose some diagnostic problem.

In achondrogenesis, limb shortening is both proximal as well as distal. Bone histopathology of achondrogenesis, like thanatophoric dwarfism, is characterized by disorganization of endochondral bone formation with lack of columnarisation of chondrocytes. The lack of matrix surrounding the chondrocytes is specific for achondrogenesis which is probably responsible for the defect in endochondral ossification(6) while the matrix is adequate in thanatophoric dwarfism.

True achondroplasia is an autosomal dominant condition and endochondral ossification is well organized and associated with orderly columns of cells in contrast to thanatophoric dwarfism.

In asphyxiating thoracic dystrophy, an autosomal recessive disorder, maximal shortening is distal. Vertebrae are normal and do not show enlargement. Polydactyly may be an associated feature in some cases.

The proper identification of the disease is more than academic curiosity since the dwarfism in infants is commonly confused with achondroplasia which is transmitted as autosomal dominant trait and therefore,

has ominous genetic implications while thanatophoric dwarfism is a reflection of dominant mutation(7).

The etiology of thanatophoric dwarfism remains unknown. In our case, mother's 'TORCH' test was positive for rubella and CMV antibodies but role of these infections in the etiology needs to be confirmed by further studies. Because of a small thoracic cage capacity and hypoplastic lungs, early respiratory distress is the cause of death in this condition.

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Typhoid Fever Before Two Years of Age

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Typhoid fever continues to remain a major public health problem in developing countries like India, causing significant morbidity. The clinical pictures of the disease in infants and young children is variable and nonspecific. The disease is usually severe and recovery is delayed(1).

There are many reports of typhoid fever in young children and the incidence below the age of five years is 13.5 to 60% of all typhoid cases in the pediatric age group(1-7). But there is hardly any information on the clinical profile and outcome of this disease in children below 2 years of age. Therefore, this study was planned to examine the clinical profile of typhoid fever in cases less than 2-years-old.

Material and Methods

Eighteen children below the age of two years, suffering from typhoid fever admitted to pediatric ward of Holy Family Hospital, New Delhi over a period of 14 months (June 1990 to July 1991) were studied and their clinical details analyzed.

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