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ANSWER: THANATOPHORIC DYSPLASIA

Figure 1a: A chest, abdomen and limbs radiograph in AP view, showing shortened and bowed long bones associated with metaphyseal flaring giving a telephone handle appearance.
Figure 1b: A chest radiograph in AP view, showing elongated narrowed chest cavity with short anterior rib and bilateral costochondral junctions appear widened.
Figure 1c: Lateral spine radiograph showing flattening of the vertebral bodies.

Thanatophoric dysplasia is a congenital skeletal anomaly which occurs in 1 in 20 000 to 50 000 newborns. It is a lethal condition. It is divided into type I and type II, with a higher incidence in type I.[1] Patients with thanatophoric dysplasia have shortening of the upper and lower limbs, redundant skin fold along with the limbs, narrow chest, flat vertebral bodies (platyspondyly) and macrocephaly.

Thanatophoric dysplasia can be suspected during antenatal sonography during second or third trimester. All femur length less than 95% confidence limits must be further investigated for short limbed dwarfism.[2] The long bones measurements are one of the crucial parameters in determining which limbs are severely shortened.[3] There are three categories of dysplasias; rhizomelic, mesomelic and micromelic dysplasia. Other associated abnormalities in the antenatal ultrasound examinations are polyhydramnios, hydrops, growth restriction, ventriculomegaly, a small thorax and flattened vertebrae.[1] There is also abnormal growth of the temporal lobe.[1] The diagnosis of thanatophoric dysplasia could not be done by antenatal ultrasound alone, it needs to be accompanied by the postnatal clinical and radiological findings.[1] The radiological findings include short, horizontal with flaring of anterior ends ribs and small scapulae. The long bones are marked by micromelia with metaphyseal flaring.

Thanatophoric dysplasia has a very poor prognosis and usually results in stillbirth or death within minutes of life. This is primarily due to respiratory distress because of pulmonary hypoplasia or brainstem compression due to underlying hydrocephalus[4]. There are only a few long-term survivals of this condition but only up to 4 to 7.5 years old. Only one case is documented that the patient lived up to late twenties[4]. However, they had severe skeletal deformity and poor cognitive function with seizure as there is malformation in the development of the cortex[4].

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