

Skeletal dysplasias and syndromes

Keith A. Kronemer and Thomas E. Herman

The approach to the skeletal dysplasias and syndromes taken here is based primarily on radiographic findings, with the addition of pertinent clinical data, including inheritance patterns. Owing to a rapidly expanding knowledge of the human genome, a chromosomal map classification of skeletal dysplasias and syndromes is increasingly valuable. However, because some osteochondrodysplasias do not yet have a confirmed genetic locus, a workable chromosomal classification of these conditions does not exist.

Achondroplasia group

The achondroplasia group can also be called the FGFR 3 disorders. These are conditions including achondroplasia with mutations in the *FGFR 3* (fibroblast growth factor receptor 3) gene located at genetic locus 4p16.

Thanatophoric dysplasia

Affected children are usually stillborn or die shortly after birth owing to hypoplastic lungs. The fetus or infant has marked short-limbed dwarfism, a large head with frontal bossing, and a depressed nasal bridge. Numerous skinfolds are present. The child has a relatively long trunk.

Radiographic findings include marked rhizomelic shortening of the long bones with metaphyseal flaring and osseous bowing and widening. Pronounced flattening of the vertebral bodies, with more constriction of their mid-portions and wide intervertebral disc spaces, is evident, giving the appearance of an inverted U or H to each vertebra on frontal radiographs. The thorax is slender, owing to short ribs with flared anterior ends. Small, rectangular iliac bones, small sacroiliac notches and short, wide pubic and ischial bones are seen (Figure 10.1).

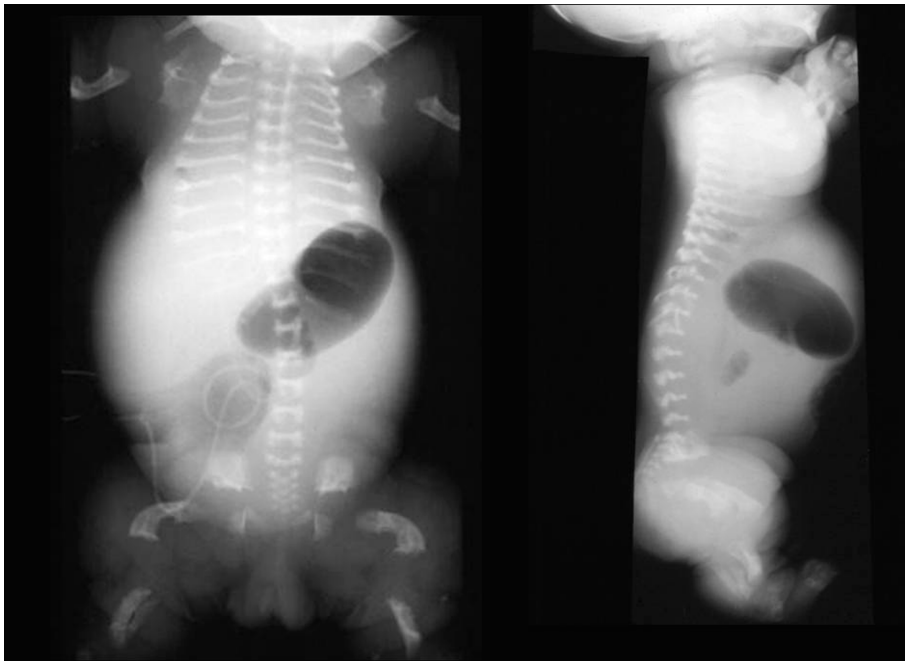


Figure 10.1 Thanatophoric dysplasia: frontal and lateral radiographs of the chest and abdomen in a stillborn infant after attempted resuscitation. Diffuse platyspondyly is present. The square iliac bones are seen with small sacroiliac notches. The femurs have a characteristic telephone-receiver appearance. Striking micromelia is present.