53 Kniest Dysplasia

Metatropic dwarfism type II, pseudometatropic dysplasia

Macrocephaly, flat face, short neck, cleft palate, bowing/shortening of long bones, prominent joints

Frequency: Rare.

Genetics

Autosomal dominant (OMIM 156550), due to mutation of the *COL2A1* at 12q 13.11–2; somatic and germinal mosaicism occur; a lethal variant, possibly autosomal recessive, is recognized (OMIM 245190).

Clinical Features

- Disproportionate dwarfism identifiable at birth
- Large head, round face, flat midface, depressed nasal bridge
- Severe myopia, cataracts, vitreoretinal degeneration, retinal detachment (40 %)
- Cleft palate (40 %)
- Hearing loss (usually develops before puberty), recurrent otitis media
- Short trunk (length may be normal at birth), short neck, sternal protrusion
- Kyphoscoliosis
- Shortening and bowing of long bones
- Prominent, stiff joints
- Clubfoot

Differential Diagnosis

- Metatropic dysplasia
- Spondyloepiphyseal dysplasia congenita
- Dyssegmental dysplasias, Rolland-Desbuquois type, and Silverman-Handmaker type

Radiographic Features

Spine

- Severe platyspondyly (especially upper thoracic spine), with anterior wedging of vertebral bodies and irregular end-plates
- Coronal clefts, lumbar (not mandatory, in infancy)
- Narrow interpediculate distance (lumbar)
- Abnormal odontoid (short, tall, wide), anterior fusion of C1–2, atlanto-occipital instability
- Cervical and dorsal kyphosis, lumbar hyperlordosis

Pelvis

- Trefoil pelvis (broad ilia with hypoplasia of basilar portions)
- Poorly ossified pubic bones
- Broad and short femoral necks, unossified capital femoral epiphyses (until adolescence in severe cases)
- Coxa vara

Limbs

- Short, slightly bowed, dumbbell-shaped long bones with flared metaphyses
- Enlarged, irregular, punctate epiphyses
- Irregular growth plates (occasionally with inverted V-shaped configuration)
- Loss of normal trabecular pattern, ground-glass appearance of bones
- Reduced joint spaces

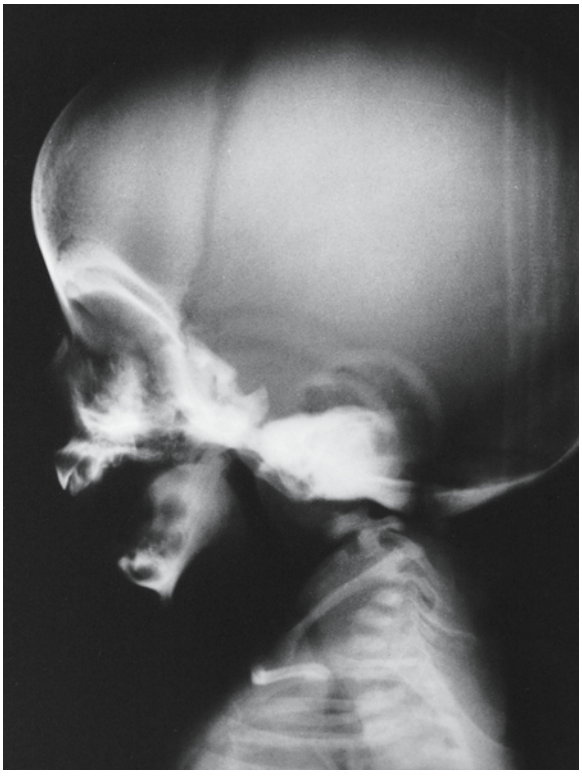

Fig. 53.1. Patient 1, age 7 months. The calvarium is enlarged, while the skull base and cranial bones are relatively small. Note striking cervical kyphosis

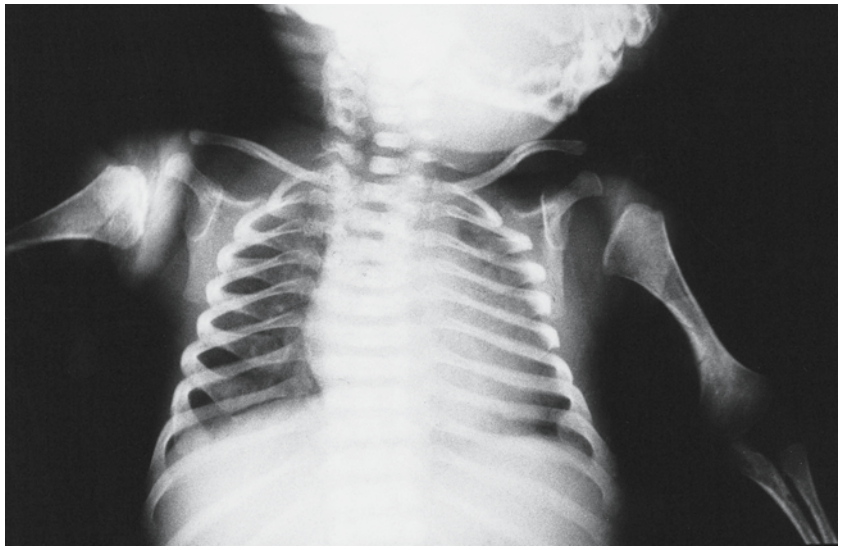

Fig. 53.2. Patient 1, age 7 months. Note short, dumbbell-shaped humeri with splayed metaphyses. The thorax is broad (in contrast to the thorax seen in metatropic dysplasia, with which Kniest dysplasia can be confused in infants)

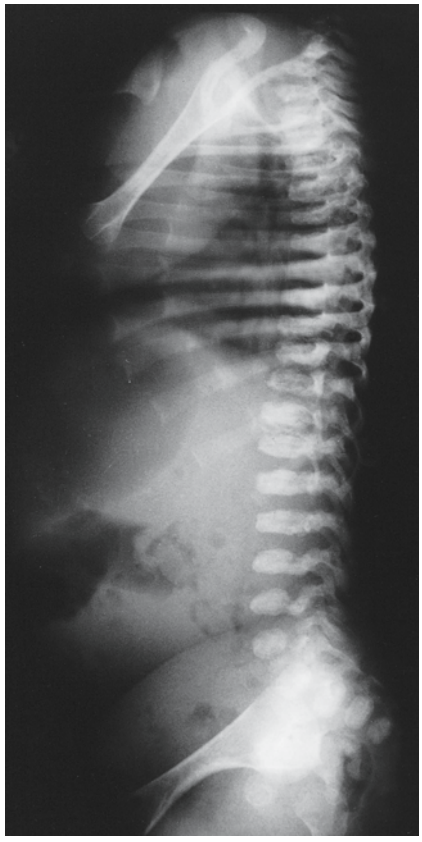

Fig. 53.3. Patient 1, age 7 months. There is generalized platyspondyly, with anterior wedging of vertebral bodies and persistent coronal clefts or end-plate notches (a remnant of an earlier cleft) in the lower lumbar spine

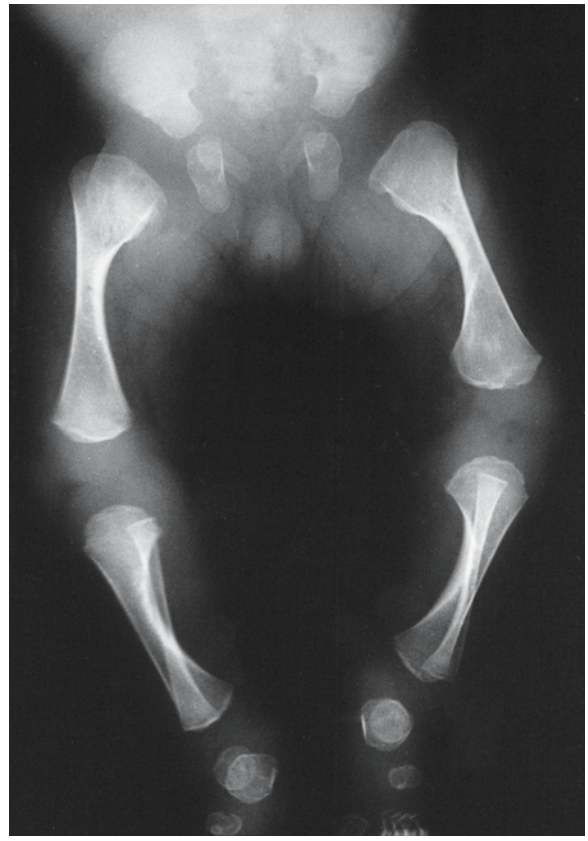

Fig. 53.4. Patient 1, age 7 months. Femurs and tibias are short, each with a striking dumbbell configuration. Tibias are mildly bent. Metaphyses are splayed and round; epiphyseal ossification centers of proximal and distal femurs and proximal tibias have not yet appeared. Pelvis shows a unique trefoil shape, with broad iliac wings, narrow basilar portions, and increased iliac angles. Ischia are broad and vertical, and pubic rami are poorly ossified

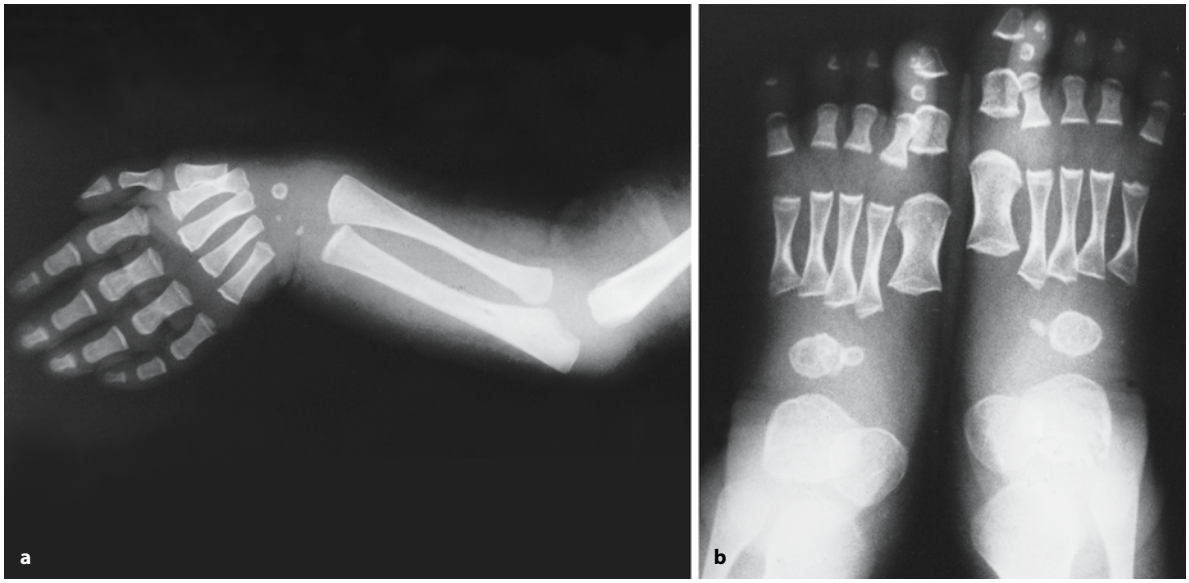

Fig. 53.5 a, b. Patient 1, age 7 months. Note short tubular bones of **a** hands and **b** feet, with wide ends. The middle phalanges of toes 3–5 are unossified

Hands

- Short tubular bones with wide ends
- Flattened and squared-off epiphyses of tubular bones
- Pseudoepiphyses
- Retarded carpal ossification, very irregular carpal bones

Skull

- Enlarged neurocranium, late-closing anterior fontanel
- Flattened cranial base, anteriorly displaced sella turcica
- Small facial bones

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