100 Spondyloepiphyseal Dysplasia Congenita

SED congenita, Spranger-Wiedemann disease

Short-trunk dwarfism, normal-sized hands and feet, chest deformity, flat face, myopia

Frequency: 1 in 100,000 births.

Genetics

Autosomal dominant (OMIM 183900), due to mutation in the *COL2A1* gene at 12q13.11–13.2; allelic mutations of the same gene also cause achondrogenesis type II, hypochondrogenesis, Kniest dysplasia, autosomal dominant spondyloarthropathy, and the COL2 type of Stickler dysplasia. Spondyloepimetaphyseal dysplasia, Strudwick type, may be the same as the disorder discussed here.

Clinical Features

- Short-trunk dwarfism, short spine, short neck
- Normal head, flat face, wide-set eyes
- Myopia (50%), retinal detachment (rare)
- Cleft palate (not mandatory), micrognathia
- Sensorineural hearing loss
- Scoliosis, kyphosis, lumbar lordosis
- Barrel chest, pectus carinatum
- Hypoplastic abdominal muscles, inguinal hernias
- Genu valgum, (less commonly genu varum)
- Short limbs, normal-sized hands and feet
- Clubfoot (not mandatory), pes planus
- Waddling gait
- Retarded motor development, hypotonia

Differential Diagnosis

- Other spondyloepiphyseal dysplasias (e.g., recessive type, Sensenbrenner type, Lie type)
- Other type II collagenopathies
- Morquio syndrome

Radiographic Features:

1. At Birth and During Infancy

Generalized Skeletal Abnormalities

- Retarded ossification of the skeleton (pubic bones, knee epiphyses, talus, and calcaneus)

Limbs

- Short tubular bones, flared metaphyses

Spine

- Absent ossification of the upper cervical vertebrae, retarded ossification of the sacrum
- Pear-shaped vertebrae (flat, with dorsal wedging)

Radiographic Features:

2. Childhood

Spine

- Flat and immature vertebral bodies with anterior ossification defects, flat and dorsally wedged vertebral bodies
- Odontoid hypoplasia, atlanto-axial instability

Pelvis

- Retarded ossification of pubic bones
- Horizontal acetabular roofs
- Retarded ossification of femoral head and neck, coxa vara

Limbs

- Varying degrees of epiphyseal and metaphyseal abnormalities
- Shortening of long bones (usually proportionate to the degree of epi- and metaphyseal involvement)
- Delayed appearance of the carpal and tarsal ossification centers

Chest

- Anteriorly splayed ribs

Radiographic Features:

3. Adulthood

Spine

- Short spine
- Moderate kyphoscoliosis, lumbar hyperlordosis
- Flattening and irregularity of the vertebral endplates
- Odontoid hypoplasia and/or lack of fusion with the body of C2

Pelvis

- Coxa vara, high-riding femoral trochanters
- Small, deformed femoral heads

Limbs

- Shortening of long bones
- Varying degrees of epiphyseal flattening and deformity
- Relatively normal bones of the hands and feet

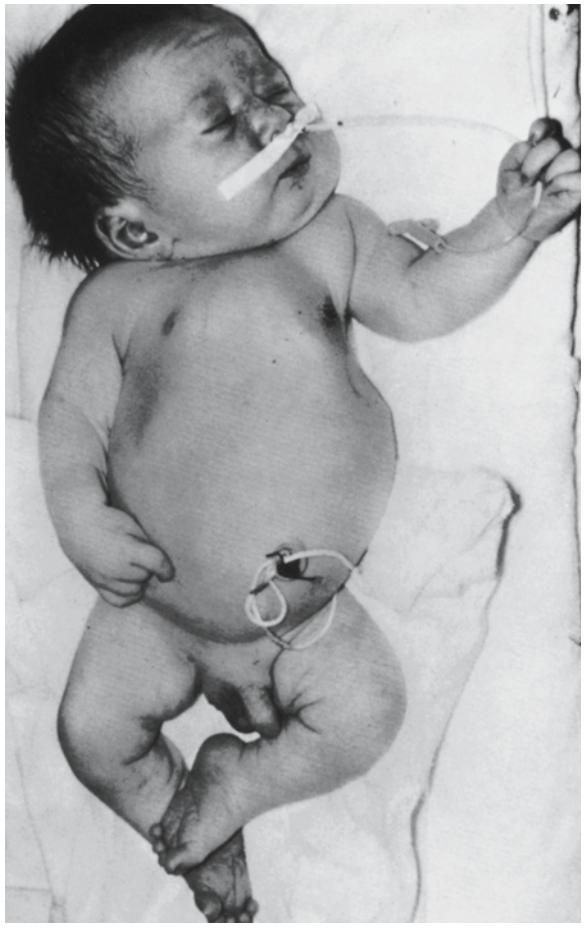

Fig. 100.1. Patient 1, newborn. Note short trunk, flat face, small deformed chest, protuberant abdomen, rhizomelic limb shortening, and bilateral clubfeet. (Reprinted, with permission, from Macpherson and Wood 1980)

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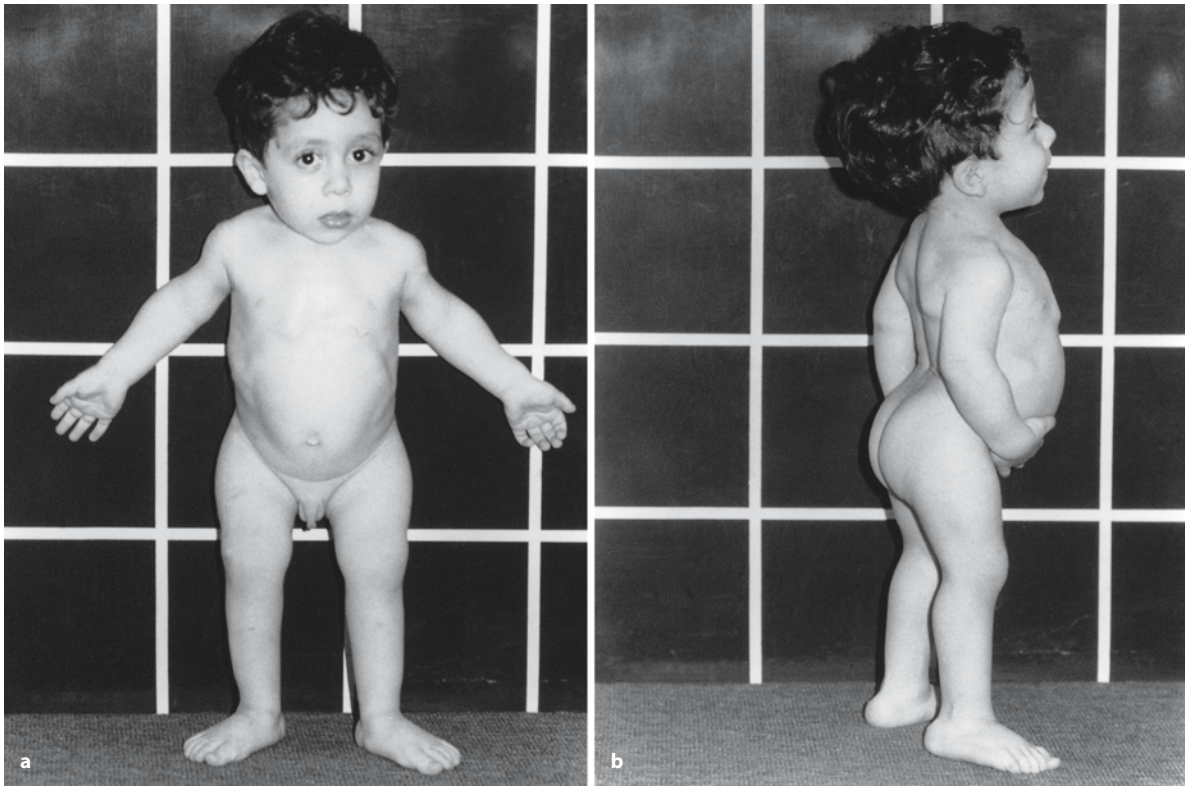

Fig. 100.2 a, b. Patient 2, age 3 years. Short-trunk dwarfism, flat face, short neck, chest deformity, lumbar hyperlordosis, normal-sized hands and feet, and flat feet. (From archive of Dr. P. Balestrazzi, University of Parma, Italy, with permission)

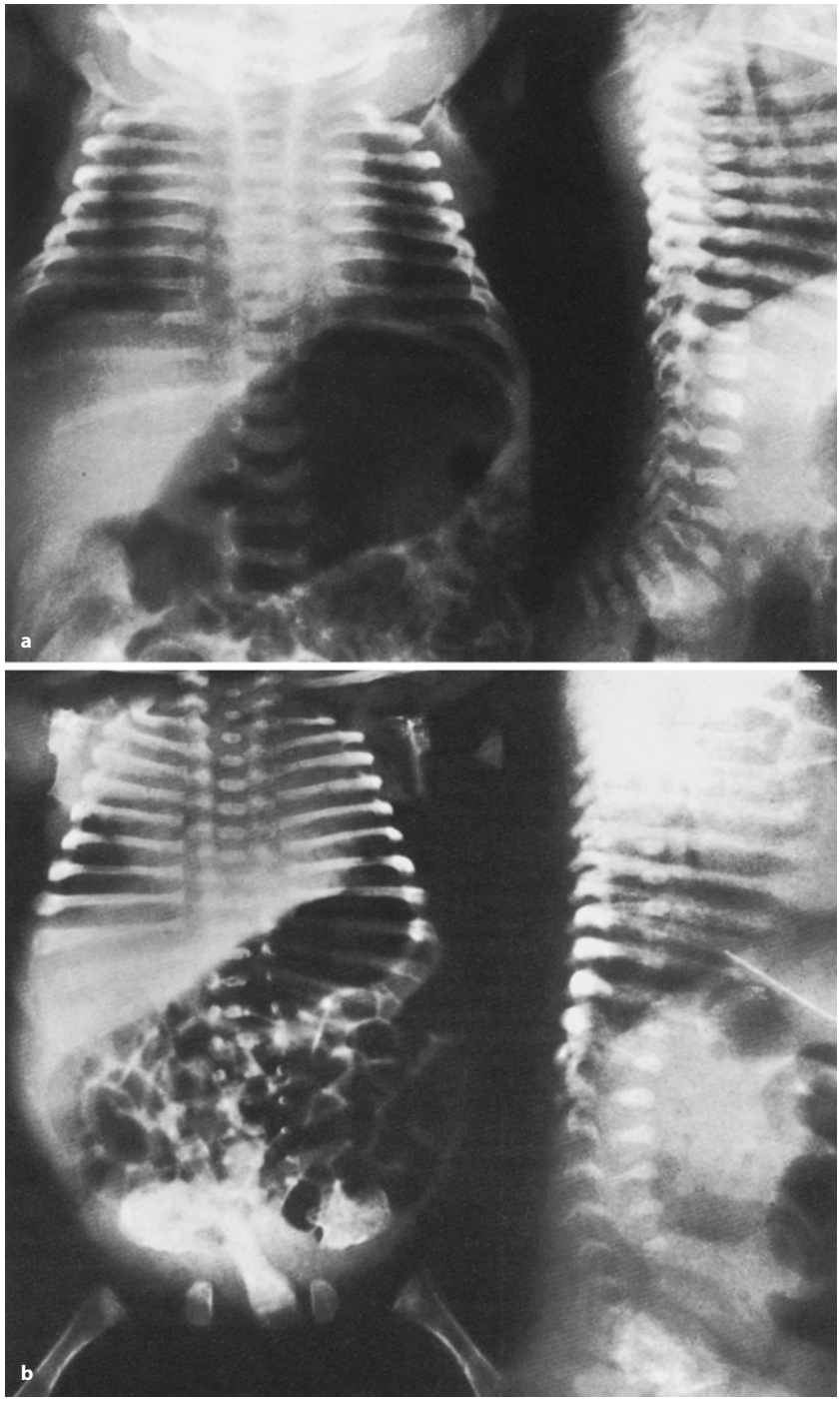

Fig. 100.3. a Patient 1, newborn; **b** patient 3, newborn. Note flat, pear-shaped vertebral bodies; small, bell-shaped thorax; absence of sternal ossification centers; and relatively squared scapulae. In patient 3 (**b**), in addition to a more severe vertebral dysplasia note hypoplastic ilia, horizontal acetabuli, absent ossification of pubic bones, and short, slightly bowed femurs. (Reprinted, with permission, from Macpherson and Wood 1980)

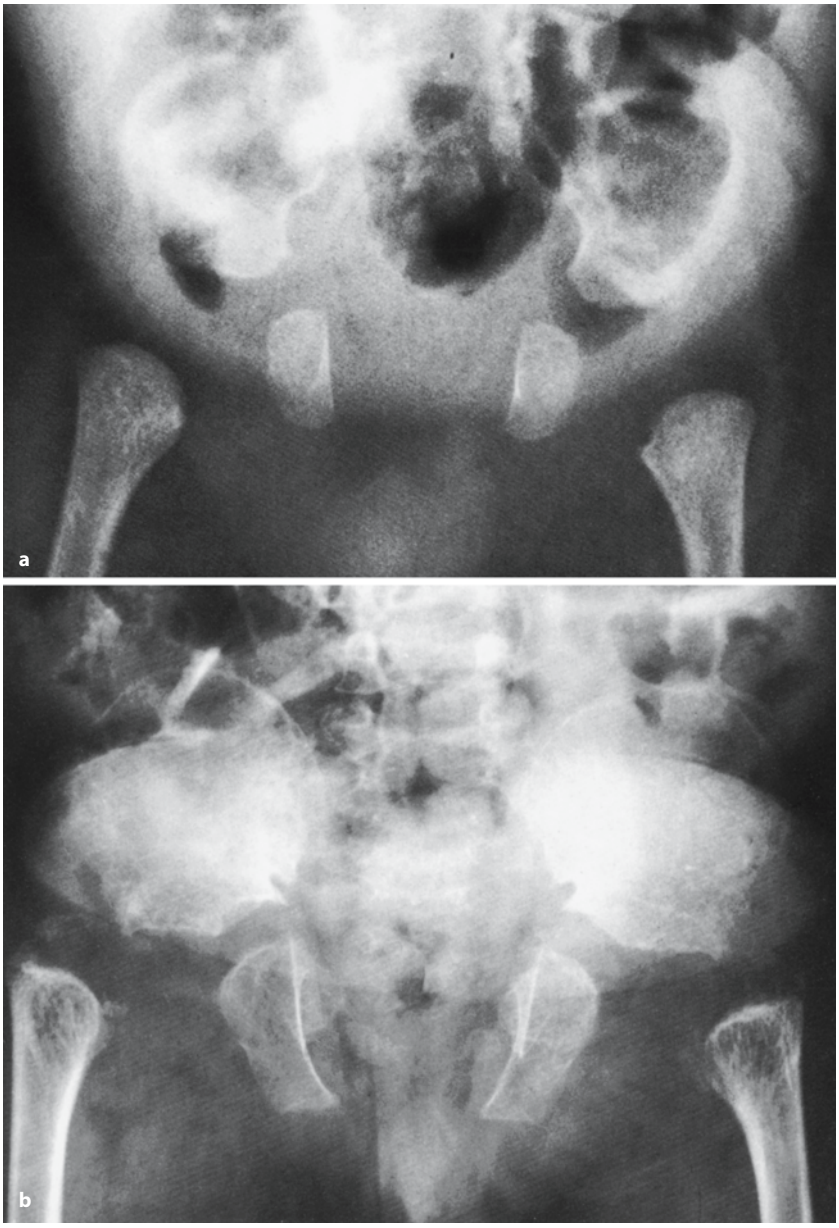

Fig. 100.4 a, b. Patient 1. **a** In neonatal phase iliac bones are hypoplastic, acetabuli are horizontal, and pubic bones are unossified. **b** At 7 years of age note hypoplastic ilia, horizontal acetabula, poorly ossified pubic rami and femoral heads, and bilateral coxa vara deformity. (Reprinted, with permission, from Macpherson and Wood 1980)

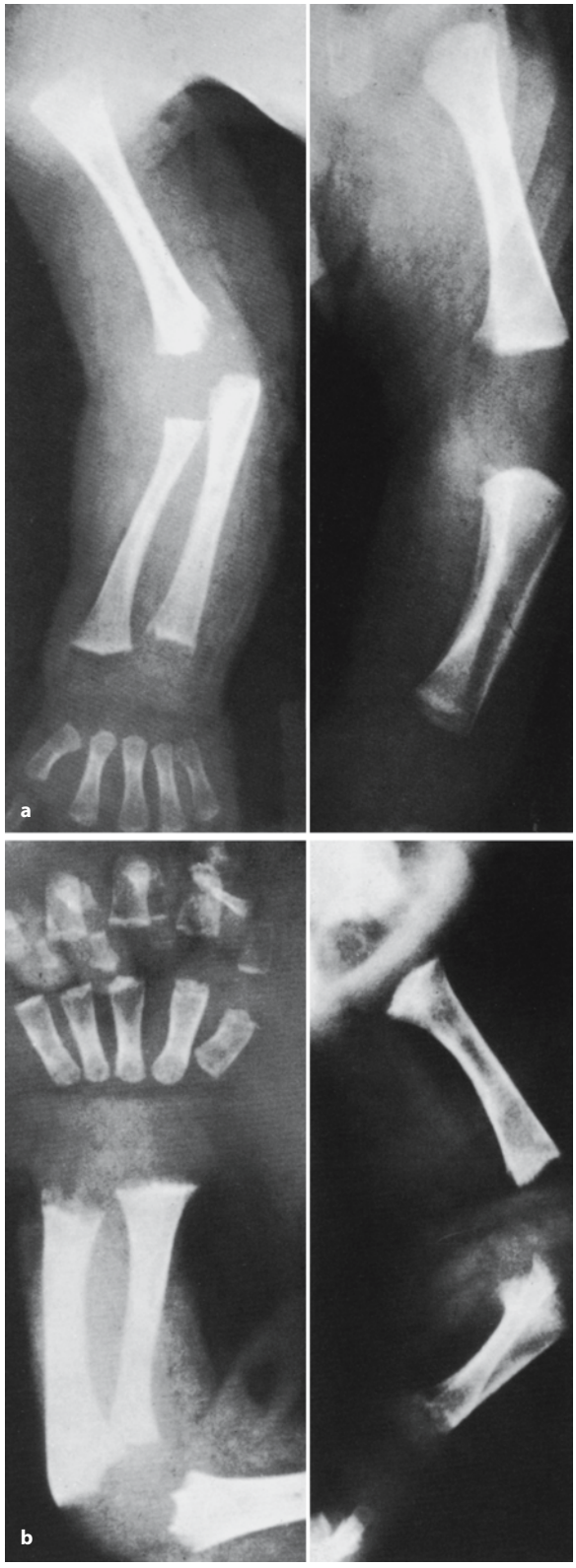

Fig. 100.5 a, b. Patients 1 (**a**) and 3 (**b**), both newborns. Note shortening of tubular bones, flaring and irregularity of metaphyses, and absent ossification of distal femoral and proximal tibial epiphyses. (Reprinted, with permission, from Macpherson and Wood 1980)

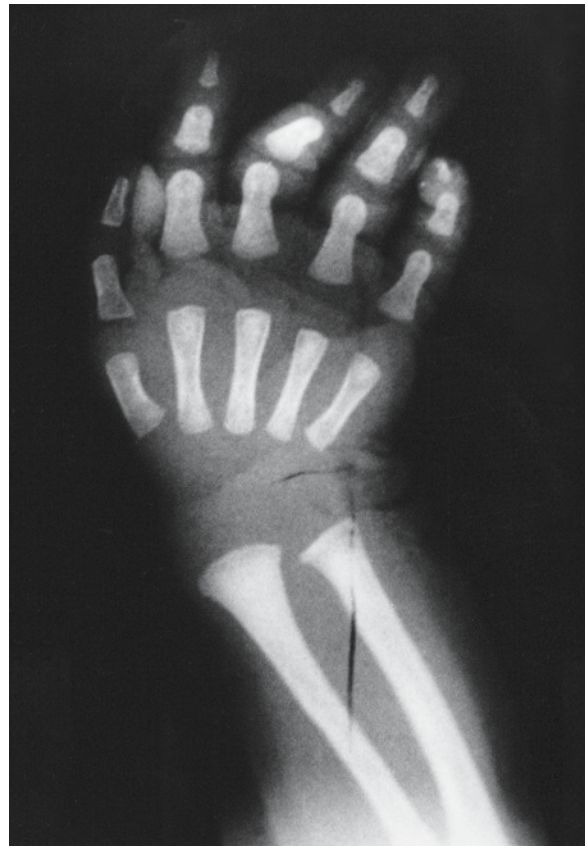

Fig. 100.6. Patient 4, newborn. Note that metacarpals and phalanges are normal in shape and size