Spondyloepiphyseal Dysplasia

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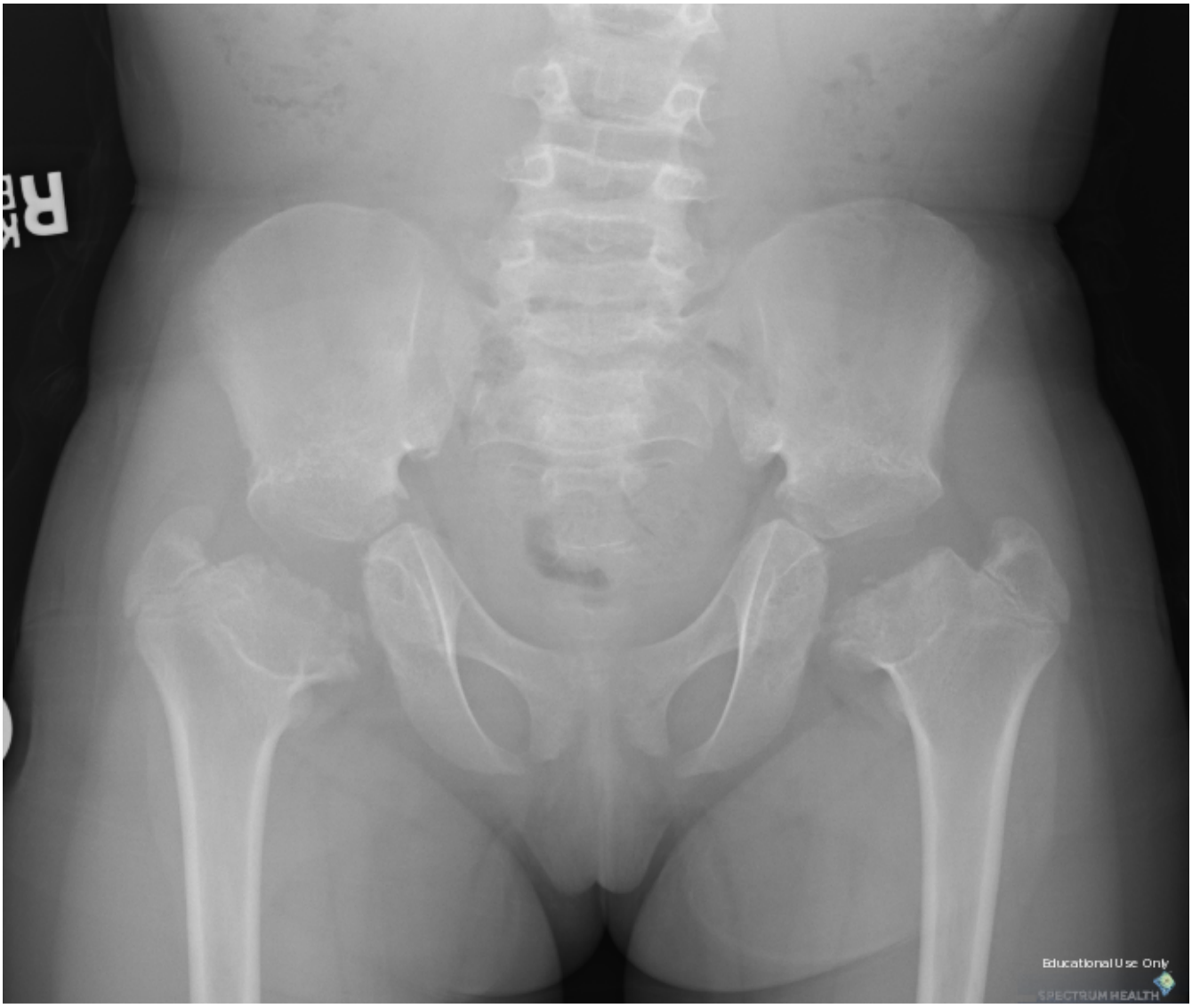
History
11 year old with short stature.

Diagnosis
Spondyloepiphyseal Dysplasia

Discussion
Spondyloepiphyseal dysplasia is an autosomal dominant dwarfism characterized by spine greater than extremities shortening, barrel chest with pectus carinatum, flat face with protruding eyes and cleft palate, genu valgus and club feet.
Radiographic findings include: 1) retarded spinal ossification in infancy, notably the cervical and sacral spine, 2) flat and/or dorsally wedged vertebral bodies often with anterior defects, 3) hypoplasia of the dens with lax ligaments, 4) short ilia and delayed ossification of the pubic bones and femoral heads with short femoral necks and coxa vara, 5) epiphyseal and metaphyseal abnormalities of the tubular bones (extremity shortness is determined by the degree of abnormality), 6) normal hands and feet although delayed appearance of the ossification centers, and 7) moderate kyphoscoliosis and lumbar lordosis.
Mutation of the COL2A1 gene is responsible. Spondyloepiphyseal dysplasia is one of several diseases related to abonormality of the COL2A1 gene and consequently bears similarities to achodrogenesis type II, hypochondrogenesis, SED congenita, autosomal dominant spondyloarthropathy and Stickler dysplasia.

Findings
CR-Short iliac bones, wide acetabula, non-ossified femoral epiphyses, short broad femoral necks in mild varus, and flat lumbar vertebrae.

Reference
Spranger JW, Brill PW, Poznanski AK. Bone dysplasias: An atlas of genetic disorders of skeletal development, 2nd Ed. Oxford University Press (2002).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