



POSNA

The Core Curriculum

Spondyloepiphyseal dysplasia (SED)

Objectives

1. Describe the two types of SED
2. Describe clinical features of each type
3. Describe the cervical spine pathology associated with SED congenita

Discussion

Although the SED's constitute a well-known group of chondrodystrophies, they are grouped with the Type II collagenopathies in the new International Group Classification, obviously acknowledging the basic defect in synthesis of type II collagen. A number of other more unusual osteochondrodysplasias are also secondary to type II collagen defects. SED is generally divided in the congenita and tarda varieties. SED congenita is obviously identifiable in the newborn; by disproportionate short stature with rhizomelic shortening, midface flattening, short neck, and barrel shaped chest. Radiographic findings include ovoid vertebral bodies and delayed epiphyseal appearance. When the epiphyses do appear, they are fragmented. Platyspondyly is characteristic of SED. The iliac bones are short and square. Hearing loss occurs in about 25%, myopia is common. Coxa vara is common. The most serious problem is atlantoaxial instability secondary to odontoid hypoplasia, os odontoideum, and/or lax ligaments. Risk of atlantoaxial instability has been related to extreme short stature and severe coxa vara. Problems can also occur in the thoracolumbar spine and paraplegia has also been linked with severe coxa vara. Other orthopaedic intervention is related to deformity correction, usually at the hip.

SED tarda is a much milder condition. SED tarda is an X-linked condition as opposed to SED congenita, which is autosomal dominant. There is usually disproportionate trunk shortening, but SED tarda is compatible with normal stature. Spinal deformity is common, including exaggerated lumbar lordosis, scoliosis, and back pain. Hip pain and/or stiffness are common presenting complaints in the second decade of life. Angular deformities of the limbs are uncommon.

References

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