POSNA

The Core Curriculum

Multiple epiphyseal dysplasia (MED)

Objectives

1. Describe clinical features suggestive of MED
2. Describe orthopaedic problems related to MED
3. Describe a method of obtaining immediate further information on the osteochondrodysplasias

Discussion

Multiple epiphyseal dysplasia is the most common of the osteochondrodysplasias. Although disorders of the skeleton have fascinated scientists and artists for centuries, the first attempt to systematically classify the osteochondrodysplasias was Sir Thomas Fairbank's atlas in 1951. Since that time, a number of efforts have been published, and constantly revised as new information becomes available, from structural, ultrastructural, and genetic research. The latest effort was by an international working group in 1997, and the emphasis was shifted from the former radiodiagnostic and morphologic criteria to a present etiopathogenetic basis. The interested student of this subject can access up to date information through the availability of On-Line Mendelian Inheritance in Man (OMIN) through the internet (http://www3.ncbi.nlm.nih.gov/Omin), and through the International Skeletal Dysplasia Web site (http://www.csms.edu/genetics/skeledys).

The multiple epiphyseal dysplasias are still recognized by that name, but do comprise several entities with intermediate forms; a milder type (Ribbing) and a more severe type (Fairbanks). A third chromosomal locus for the multiple epiphyseal dysplasias was recently located. The transmission is autosomal dominant. Patients with MED may be of normal or short stature, radiographic findings in the wrists and hands have been found to correlate with stature. The hips are most often involved. The epiphyses of the long bones appear late and present an irregular, mottled, flattened appearance. Genu varum or valgum is common. The spine is minimally involved.

From an orthopaedic perspective, children with MED may present as having mild delay in walking, stiffness, or pain, pain usually does not occur until the second decade. The femoral epiphyseal radiographic changes may be confused with Legg-Perthes disease. Realignment procedures of the hip or knee may be of benefit in better distributing weightbearing forces. Arthroplasty is the only option for late cases with established degenerative changes.

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