Spondyloepimeta physeal Dysplasia with Multiple Dislocations

Alternative Names
SEMD-MD
Spondyloepimeta physeal Dysplasia with Multiple Dislocations, Leptodactylic Type
Spondyloepimeta physeal Dysplasia with Multiple Dislocations, Hall Type

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
603546

Mode of Inheritance
Autosomal dominant

Description
The spondyloepimeta physeal dysplasias (SEMD) are a genetically heterogeneous group of conditions characterized by radiological abnormalities of the spine, epiphyses and metaphyses. The term encompasses a large group of disorders with varying phenotypes. SEMD with multiple dislocations (Hall type) has recently been demarcated as a separate disorder. The disease is characterized by striking epiphyseal and metaphyseal changes in the long bones, short stature, joint dislocations and/or laxity, carpal and hand-epiphyseal ossification delay, gracile metacarpals, mild scoliosis, and dysmorphic features such as a short and upturned nose with a depressed nasal bridge and midface hypoplasia. Ossification delay leads to narrow, curved and tapered femoral neck. Small irregular epiphyses and striations in the adjacent metaphyses are characteristic of the disease. Laryngeal stenosis or tracheomalacia may be seen in childhood. Treatment of the disorder involves maintaining mobility of the joints, and avoiding dislocations.

Molecular Genetics
SEMD-MD has been reported to be genetically transmitted in at least four different families. The disorder shows an autosomal dominant mode of inheritance.

Epidemiology in the Arab World

Iraq
Shohat et al. (1993) reported a distinct type of spondyloepimeta physeal dysplasia seen in two sibs and their second cousin, characterized by early onset severe short stature, small chest, and distended abdomen. They had short neck, severe lumbar lordosis, and marked genu varum due to fibular overgrowth and joint laxity. Radiography demonstrated that the patients had platyspondyly, initially noted during the first years of life, with central hypoplasia of the vertebral bodies. At a later age, the vertebra appear squared with mild interpedicular narrowing. The long bone changes, which at early age resemble those seen in achondroplasia, later include general metaphyseal irregularities and significant epiphyseal ossification delay. Shohat et al. (1993) suggested that their patients represent a previously undescribed form of spondyloepimeta physeal dysplasia, most probably transmitted as an autosomal recessive tract.

Lebanon
Megarbane et al. (2003) reported the case of a 6-year old boy, born to non-consanguineous healthy parents, who was presented to the clinic for short stature. All his siblings were normal. He had earlier suffered from hip dislocation at the age of 3-months, and his developmental milestones had been delayed. Physical examination showed his height and weight to be below the 3rd centile. He also showed relative macrocephaly, slightly low set ears, a short neck, broad thorax, brachydactyly, hyperextensible joints, a genu valgum, and flat feet. Lengths of his upper arm, forearm, hand, total lower limb, and feet were below the 3rd
centile. Radiological results showed severe delay in ossification of the epiphyses and the carpal bones, thoracic scoliosis, right hip dislocation, small and irregular epiphyses, slender femoral neck with flattened epiphyses, narrow interpedicular distances in the lower limb, and slender metacarpals, with no epiphyseal or carpal ossification. Megarbane et al. (2003) considered sponastrime dysplasia and SEMD with joint laxity as possible diagnosis, before positively identifying the boy with SEMD-MD.

References


Contributors
Pratibha Nair: 4.7.2006