Spondyloepiphysseal Dysplasia, Maroteaux Type

**Alternative Names**
SED, Maroteaux Type
Spondyloepimetaphyseal Dysplasia, Maroteaux Type
SEMD, Maroteaux Type
Pseudo-Morquio Type II Syndrome

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

**OMIM Number**
184095

**Mode of Inheritance**
Autosomal dominant

**Description**
Spondyloepiphysseal dysplasia (SED) of Maroteaux is a very rare skeletal dysplasia. Disease manifestations are restricted to the musculoskeletal system and include abnormalities of the spine, epiphyses and metaphyses and multiple dislocations of the large joints. Spondylar dysplasia, epiphyseal dysplasia of the large joints, and type E-like brachydactyly are observed with SED Maroteaux type. These abnormalities result in a short-trunk disproportionate dwarfism. Also, dysmorphic features are noticed including a short and upturned nose with a depressed nasal bridge and midface hypoplasia.

**Molecular Genetics**
Spondyloepiphysseal dysplasia (SED) Maroteaux type is inherited as an autosomal dominant pattern. To date, no specific gene has been identified to be associated with the disease.

**Epidemiology in the Arab World**

**Lebanon**
Megarbane et al. (2004) reported an 11-year-old girl who presented the features of spondyloepimetaphyseal dysplasia (SEMD) of Maroteaux. She was the only child of healthy non-consanguineous parents. She appeared normal at birth. At the age of four months, the child was noted to have dorso-lumbar kyphoscoliosis, generalized platyspondyly, small epiphyses, enlarged and flared metaphyses, and delayed bone age. She was treated by a cast until age one year when it was replaced by a corset. Her growth chart showed a marked delay when she was 18 months. She was first seen at 11 years of age for short stature. She had normal intelligence. Physical examination was performed showing a head positioned in hyperextension, a mild arched palate, prominent joints, limited elbow movements, hyperextensible wrists and fingers, brachydactyly, broad thorax, a pectus carinatum, a short trunk, a genu valgum, and flat feet. Radiological examination revealed decreased bone density, flat and enlarged vertebral bodies with sclerosis of the endplates, small and square iliac wings, short femoral neck with flattened epiphyses, flared metaphyses, and dysplastic carpal bones. The epiphyses of the knees were dysplastic, small, flattened, irregular, and fragmented. Chromosome studies were normal (46,XX). The patient had cervical spine instability and distal joint laxity. These two features were not described previously in patients with the same disorder; therefore, Megarbane et al. (2004) suggested that SEMD of Maroteaux might show clinical or genetic heterogeneity.

**References**

**Contributors**
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