Brachyolmia, Maroteaux type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Brachyolmia, Maroteaux type. ORPHA:93302

Autosomal recessive brachyolmia, Maroteaux type is a relatively mild form of brachyolmia (see this term), a group of rare genetic skeletal disorders, characterized by short trunk/short stature, generalized platyspondyly and rounding of vertebral bodies. It remains unknown whether the phenotype represents a single disease entity or a heterogeneous group of mild skeletal dysplasias.