Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria. ORPHA:99646

Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria is an extremely rare genetic disorder characterized by the unique association of enchondromatosis with D-2 hydroxyglutaric aciduria (see these terms). Clinical features include enchondromatosis (with short stature, severe metaphyseal dysplasia and mild vertebral involvement), elevated levels of urinary 2-hydroxyglutaric acid and mild developmental delay.