

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Kod ORPHA

99646

Kod OMIM

614875

Kod ICD10

Q78.8

Kod ICD11

LD24.2Y

*Źródło

orphanet