Opis choroby *

Definicja

A rare disorder characterized by epiphyseal stippling and osteoclastic overactivity. It has been described in less than 10 patients but may be underdiagnosed. It is characterized radiographically by severe stippling of the lower spine and long bones, and periosteal cloaking. Patients also have short metacarpals. The syndrome may be inherited as an autosomal recessive trait. This disorder should be included in the differential diagnosis of mucolipidosis type II. In order to make a definitive diagnosis, lysosomal storage should be investigated by electron microscopy, or enzyme assays should be performed. Familial recurrence can be easily detected by prenatal ultrasonography. This skeletal dysplasia is lethal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Pacman dysplasia
Zespół kropkowatych nasad - hiperplazja osteoklastyczna

Kod ORPHA
1952

Kod OMIM
167220

Kod ICD10
Q77.8

Kod ICD11
FB86.2

---

*Źródło

orphanet