

Dysplazja SPONASTRIME

Kod Orpha: 93357 Kod OMIM: 271510

Opis choroby *

Definicja

A rare, genetic, spondyloepimetaphyseal dysplasia disease characterized by short-limbed short stature (more pronounced in lower limbs) associated with characteristic facial dysmorphism (i.e. relative macrocephaly, frontal bossing, midface hypoplasia, depressed nasal root, small upturned nose, prognathism) and abnormal radiological findings, which include abnormal vertebral bodies (particularly in the lumbar region), striated metaphyses, generalized mild osteoporosis, and delayed ossification of the carpal bones. Progressive coxa vara, short dental roots, hypogammaglobulinemia and cataracts may be occasionally associated.

Dane

Klasyfikacja

Choroba

Synonimy

Spondylar and nasal changes with striations of the metaphyses (SPONASTRIME) dysplasia
Dysplazja kręgowo-nasadowo-przynasadowa,
typ Sponastrime
Spondyloepimetaphyseal dysplasia,
Sponastrime type

Kod ORPHA

93357

Kod OMIM

271510

Kod ICD10

Q77.7

Kod ICD11

LD24.3

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Orphanet - internetowa baza danych dotyczących rzadkich chorób i sierochych leków. ©INSERM 1999 -
Dostępna na stronie www.orphanet.pl