Opis choroby *

Definicja

Spondyloenchondrodysplasia (SPENCD) is a very rare genetic skeletal dysplasia characterized clinically by skeletal anomalies (short stature, platyspondyly, short broad ilia) and enchondromas in the long bones or pelvis. SPENCD may have a heterogeneous clinical spectrum with neurological involvement (spasticity, mental retardation and cerebral calcifications) or autoimmune manifestations, such as immune thrombocytopenic purpura, systemic lupus erythematosus (see these terms) hemolytic anemia and thyroiditis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SPENCD
Spondyloenchondromatoza
Spondylometaphyseal dysplasia with enchondromatous changes
SPENCD
Spondyloenchondromatosis
Spondylometaphyseal dysplasia with enchondromatous changes

Kod ORPHA
1855

Kod OMIM
607944

Kod ICD10
Q77.7

Kod ICD11
LD24.3

---

*Źródło

orphanet