Opis choroby *

Definicja

A rare primary bone dysplasia characterized by the association of spondylometaphyseal dysplasia, generalized joint laxity, and dentinogenesis imperfecta. Main skeletal abnormalities comprise short stature, narrow chest, scoliosis, mesomelic limb shortening, and brachydactyly. Radiographic features include severe metaphyseal irregularities of the tubular bones, platyspondyly with coronal clefts, cone-shaped epiphyses of the hands, square iliac wings, and coxa valga. Additional extraskeletal manifestations like pulmonary hypoplasia, cystic renal disease, and non-obstructive hydrocephalus have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Chondrodysplasia-dentinogenesis imperfecta-joint laxity syndrome
Chondrodysplazja - dentinogenesis imperfecta - wiotkość stawów
Chondrodysplazja Goldblatta
Dysplazja zębowo-chrzęstna
ODCD
Goldblatt chondrodysplasia
Goldblatt syndrome
ODCD

Kod ORPHA
166272

Kod OMIM
184260

Kod ICD10
Q78.8

Kod ICD11
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*Źródło

orphanet