

Opis choroby *

Definicja

CHST3-related skeletal dysplasia is a very rare bone disorder characterized clinically by short stature of prenatal onset; dislocation of the knees, hips or elbows; club feet; limitation of range of motion of large joints; progressive kyphosis; and occasional scoliosis. In a few patients, minor heart valve dysplasia has also been described. Intellect, vision and hearing are normal.

Dane

Klasyfikacja

Choroba

Synonimy

Chondrodysplasia with congenital joint dislocations, CHST3 type

Chondrodysplazja z wrodzonym zwichnięciem stawów, typu CHST3

Dysplazja kręgosłupowo-nasadowa z wrodzonym zwichnięciem stawów, typu CHST3

SDCD, CHST3 type

Spondyloepiphyseal dysplasia with congenital joint dyslocations, CHST3 type

Kod ORPHA

263463

Kod OMIM

143095

Kod ICD10

Q74.8

Kod ICD11

LD24.3

*Źródło

orphanet