

Opis choroby *

Definicja

Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis is a type 2 collagen-related bone disorder characterized by precocious, generalized osteoarthritis (with onset as early as childhood) and mild, dysplastic spinal changes (flattening of vertebrae, irregular endplates and wedge-shaped deformities) resulting in a mildly short trunk.

Dane

Klasyfikacja

Choroba

Kod ORPHA

93279

Kod OMIM

604864

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet