

Opis choroby *

Definicja

Geroderma osteodysplastica (GO) is characterized by lax and wrinkled skin (especially on the dorsum of the hands and feet and abdomen), progeroid features, hip dislocation, joint laxity, severe short stature/dwarfism, severe osteoporosis, vertebral abnormalities and spontaneous fractures, and developmental delay and mild intellectual deficit.

Dane

Klasyfikacja

Zespół wad wrodzonych Karłowatość Walta Disneya

Kod ORPHA

2078

Kod OMIM

231070

Kod ICD10

Q82.8

Kod ICD11

LD2B

*Źródło

orphanet