Opis choroby *

Definicja

A rare genetic disease characterized by infantile onset of recurrent skin ulcerations, arthralgias, fever, peri-articular fistulous osteolysis, oligodontia, nail dystrophy, and keratitis. The disease takes a self-limiting course in childhood but results in severe cicatrization, chronic arthroses, pseudoacromegalic appearance of hands and feet, secondary scoliosis, and visual impairment. There have been no further descriptions in the literature since 1983.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1657

Kod OMIM
221810

Kod ICD10
Q82.8

Kod ICD11
LD27.0Y

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*Źródło

orphanet