

Opis choroby *

Definicja

Spondyloepiphyseal dysplasia (SED), MacDermot type is characterized by short stature, femoral epiphyseal dysplasia, mild vertebral changes and sensorineural deafness.

Dane

Klasyfikacja

Zespół wad wrodzonych Spondyloepiphyseal dysplasia-myopia-sensorineural deafness syndrome
Spondyloepiphyseal dysplasia - myopia - sensorineural deafness
Spondyloepiphyseal dysplasia-myopia-sensorineural hearing loss syndrome

Synonimy

Kod ORPHA

163668

Kod OMIM

184000

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet