

Opis choroby *

Definicja

Spondyloepimetaphyseal dysplasia, aggrecan type is a new form of skeletal dysplasia characterized by severe short stature, facial dysmorphism and characteristic radiographic findings.

Dane

Klasyfikacja

Choroba

Synonimy

SEMD, aggrecan type

SEMD, typ agrekan

Kod ORPHA

171866

Kod OMIM

612813

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet