

Opis choroby *

Definicja

Dyssegmental dysplasia, Silverman-Handmaker type is a rare, genetic, primary bone dysplasia disorder, and lethal form of neonatal short-limbed dwarfism, characterized by anisospondyly, severe short stature and limb shortening, metaphyseal flaring and distinct dysmorphic features (i.e. flat facial appearance, abnormal ears, short neck, narrow thorax). Additional features may include other skeletal findings (e.g. joint contractures, bowed limbs, talipes equinovarus) and urogenital and cardiovascular abnormalities.

Dane

Klasyfikacja

Choroba

Kod ORPHA

1865

Kod OMIM

224410

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet